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**An investigation into the experiences of  
mixed ancestry parents with a  
preschool child with Down syndrome**



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**SUBMITTED TO THE UNIVERSITY OF CAPE TOWN**  
In partial fulfilment of the requirements for the degree  
**MSC (MED) GENETIC COUNSELLING**

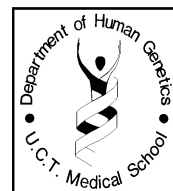
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## **DECLARATION**

I, Chantelle Jennifer Scott, hereby declare that the work on which this dissertation is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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## ABSTRACT

Down syndrome (DS) is the most common chromosomal cause of intellectual disability and in the South African (SA) mixed ancestry population a prevalence of 1.54 in 1 000 live births have been reported. Children with DS have a unique variety of health concerns and therefore regularly need to be followed-up. In the Western Cape (WC), the Red Cross War Memorial Children's Hospital (RCWMCH) and Tygerberg Hospital (TBH) have specialised services that are offered to children with DS and their parents. The majority (53.9%) of the WC population consists of the mixed ancestry group. The aims of the current study were to investigate the knowledge regarding DS and the needs of parents of mixed ancestry with a preschool child with DS; to explore the extent to which needs are being met and to identify the problems these parents experience.

A qualitative phenomenological approach was selected as it aims at describing participants' understanding of experiences and behaviour, and meanings they attach to these. Ten semi-structured interviews were conducted with mixed ancestry parents with a child with DS. These interviews were conducted in the participant's language of choice and signed informed consent was obtained prior to the interview.

The majority of the participants had a good basic knowledge of the characteristics. Most of them knew that DS is caused by a genetic anomaly, but could not recall that it was due to extra material from chromosome 21. The level of understanding did not seem to be related to the level of income or education of the participants. However, an increased level of understanding was associated with increased personal interactions with individuals with DS. The majority of the participants were satisfied with the health care and supportive services at the RCWMCH and TBH and felt that their needs were being adequately addressed. The difficulties parents experienced were mainly related to lack of support from their partner, frequent illness of their child with DS, as well as financial and transport problems. Even though the participants described an initial reaction of shock when they heard about their child's diagnosis, the majority felt that their child had had a positive impact on their lives and that of their family. Only half of the participants believed that it was possible to have another child with DS and the majority lacked an accurate understanding of their recurrence risks. Overall the participants felt positively about the available prenatal screening and diagnostic testing options. However none of them would opt to terminate a pregnancy if the foetus has DS.

This was the first study of its kind in SA conducted amongst the mixed ancestry group in the Cape Town area. The findings of this study will enhance the understanding of health care professionals regarding the experiences of these parents and the difficulties in their daily lives. An increased sensitivity will hopefully lead to an increase of the successful adaptation of these parents to their child with DS.

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## LIST OF TERMS AND ABBREVIATIONS

<b>AAP</b>	American Association of Paediatrics
<b>AFP</b>	Alpha feta protein
<b>AMA</b>	Advanced maternal age
<b>Atresia</b>	A condition in which an opening or passage for the tracts of the body is absent or closed.
<b>BF</b>	Biopsy forceps
<b>Brachycephalic</b>	Back of head flattened.
<b>Coeliac disease</b>	Autoimmune disorder of the small intestine that occurs in genetically predisposed people of all ages from middle infancy onwards.
<b>Cor pulmonale</b>	Acute strain or hypertrophy of the right ventricle caused by a disorder of the lungs or of the pulmonary blood vessel.
<b>CVS</b>	Chorionic villus sampling
<b>CXR</b>	Chest X-ray
<b>DS</b>	Down Syndrome
<b>DSA</b>	Down Syndrome Association

<b>DSAWC</b>	Down Syndrome Association Western Cape
<b>ECG</b>	Electrocardiogram
<b>ENT</b>	Ear, nose and throat
<b>Exomphalos</b>	A midline defect with herniation of abdominal contents into the base of the umbilical cord, confined by an amnioperitoneal membrane.
<b>Fβ-hCG</b>	Free beta subunit of human chorionic gonadotrophin
<b>FISH</b>	Fluorescent in situ hybridisation
<b>Gait</b>	A particular way or manner of moving on feet.
<b>Glaucoma</b>	Any of a group of eye diseases characterised by abnormally high intraocular fluid pressure, damaged optic disk, hardening of the eyeball, and partial to complete loss of vision.
<b>hCG</b>	Human chorionic gonadotrophin
<b>Hirschsprung's disease</b>	An abnormality in which certain nerve fibres are absent in segments of bowel, resulting in severe bowel obstruction.
<b>ID</b>	Intellectual disability
<b>Karyotype</b>	The characterisation of the chromosomal complement of an individual, including number, form and size of the chromosomes.

<b>Mosaicism</b>	A condition in which tissues of genetically different types occur in the same organism.
<b>NA</b>	Not applicable
<b>No</b>	Number
<b>NT</b>	Nuchal translucency
<b>NTD</b>	Neural tube defect
<b>NSGC</b>	National Society of Genetic Counselors
<b>P</b>	Participant number
<b>PAPP-A</b>	Plasma protein-A
<b>R</b>	Rand
<b>RCWMCH</b>	Red Cross War Memorial Children's Hospital
<b>Robertsonian translocation</b>	A type of chromosome rearrangement that is formed by the fusion of the whole long arms of two acrocentric chromosomes (chromosomes with the centromere very near the end of the arm).
<b>Stenosis</b>	A constriction or narrowing of a duct or passage.
<b>T21</b>	Trisomy 21

<b>TA</b>	Transabdominal
<b>TBH</b>	Tygerberg Hospital
<b>TC</b>	Transcervical catheter
<b>TOP</b>	Termination of pregnancy
<b>Toy Library</b>	Preventive service, filling gaps in existing provision for all families with babies and young children and for people with special needs. By offering a supportive service to parents and by making available and lending appropriate toys, they extend the opportunity for shared play into the home.
<b>UK</b>	United Kingdom
<b>USA</b>	United States of America
<b>WC</b>	Western Cape

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# Chapter 1

## INTRODUCTION





## 1.1 INTRODUCTION

Down syndrome (DS) is the most common chromosomal cause of intellectual disability and results from either a partial or complete triplication of chromosome 21 (Bittles et al 2007). The worldwide prevalence of DS is reported to be 1 in 650-1000 live births. These figures vary between countries because of differences in maternal age, access to antenatal diagnosis, and social attitudes towards termination of pregnancy (Roizen and Patterson 2003; Bittles et al 2007). In South Africa (SA) the prevalence has been documented as being between 1.8 and 2.01 per 1000 live births (Christianson 1996). The overall birth prevalence of DS in Cape Town, SA, was reported as being 1.49 per 1000 between 1974 and 1993. The prevalence rate in the caucasian, mixed ancestry and black population groups were reported as 1.88, 1.54 and 1.29 per 1000 respectively (Molteno et al 1997).

Children with DS frequently present with many medical conditions and a variety of congenital malformations and therefore it is essential for them to have routine assessments and ongoing medical management (Marder and Dennis 1997; Van Cleve and Cohen 2006; Lampret and Christianson 2007). In the Western Cape (WC) these children are mainly referred to The Red Cross War Memorial Children's Hospital (RCWMCH) (Figure 1.1) or Tygerberg Hospital (TBH) (Figure 1.2). Their diagnosis is confirmed by a paediatrician or medical geneticist after which they are referred to the genetic counselling services. Their health and management is followed up by the genetic services for their first year of life, after which the developmental clinics take over until school-going age.



**Figure 1.1: RCWMCH (<http://myhopefortheflowers.blogspot.com>)**



**Figure 1.2: TBH (<http://jhsph.edu/iiru/news.html>)**

When the news is broken to parents that their child has DS they immediately start experiencing the loss of having a healthy, normal developing baby and they start worrying about what the future holds for their child, about the relationships their child will have with others and even about the occupation their child will have as an adult (Kidder and Skotko 2001). This experience in itself is very anxiety-provoking. Furthermore because of the unique health and developmental concerns and additional ongoing challenges associated with raising a child with DS, families of these children experience higher levels of stress than families of typically developing children (Lam and McKenzie 2002, Van Riper 2007). For these parents to cope with the ongoing stress it is vital for them to believe that their child is receiving the appropriate health care services (Wolf et al 1989). The perception, that they are receiving adequate social support for themselves and their child promotes successful family adaptation (Bristol 1984).

To date few studies have focused on parents' experiences of having a child with DS and the supportive services they are offered. Research has been done in first world countries like the United Kingdom (UK), United States of America (USA) and Australia; on how to improve the health care and supportive services, but these models are not always appropriate or feasible within the very different socio-economic and multi-cultural context of developing countries like SA. Furthermore, few studies have focused on the self-reported needs of parents and which needs they felt were being inadequately addressed (Freedman and Boyer 2000). In SA there is a paucity of studies focusing on the experiences and self-reported needs of parents with children with DS (Christianson and Modell 2004; Lampret and Christianson 2007). Due to this there are no available reports of what is known and what is not known about the experiences of parents of a child with DS in SA. In order to promote successful adaptation of parents of a child with DS, it is essential to explore these needs in order to provide genetic counsellors and other health care professionals with insight into the experiences of these parents and to help improve the available health care and supportive services.

According to the National Census of 2001, the majority (53.9%) of the WC's population consists of individuals from the mixed ancestry or better known as the coloured group (Lehohle 2004). This population group developed from the mixed offspring of the Malay Slaves and the Dutch settlers and soldiers in the 1600s and admixture with the indigenous populations, like the Khoikhoi, San and later the African Xhosa tribe (Attlee 1947). Thirty-six percent of individuals in the mixed ancestry group have either only education on a primary level or have had no schooling. Only 17.7% of this population group have completed education on a secondary level. The poverty rate amongst this group is 19.2% and 11.3% of the working aged group are employed (Lehohle 2004). The mixed ancestry population group of the WC was selected as the focus point of the current research as they constitute the majority of the population and it is assumed that they have more needs due to their lower-educational backgrounds and lower socio-economic status.

### 1.2 AIMS

The aims of this study were to:

- investigate the level of knowledge regarding DS and the needs of parents of mixed ancestry with a preschool child with this condition;
- explore the extent to which these needs are being met by the available health care and supportive services; and
- identify the problems experienced by these families with a preschool child with DS.

### 1.3 OBJECTIVES OF STUDY

The objectives of the research study were to:

- describe the sociodemographic profile of the participants;
- describe the experiences of the parents at the time of the delivery of the news regarding their child's diagnosis;
- investigate the participants' level of satisfaction with the counselling services;
- describe the impact of the child's diagnosis on the parents and their families;

- establish how the parents have been coping since the diagnosis;
- determine how having a child with DS has impacted on the family;
- measure the parents' level of knowledge of DS; and
- investigate their attitudes towards available prenatal screening and diagnostic testing for future pregnancies.

#### **1.4 OUTLINE OF RESEARCH DESIGN AND METHODOLOGY**

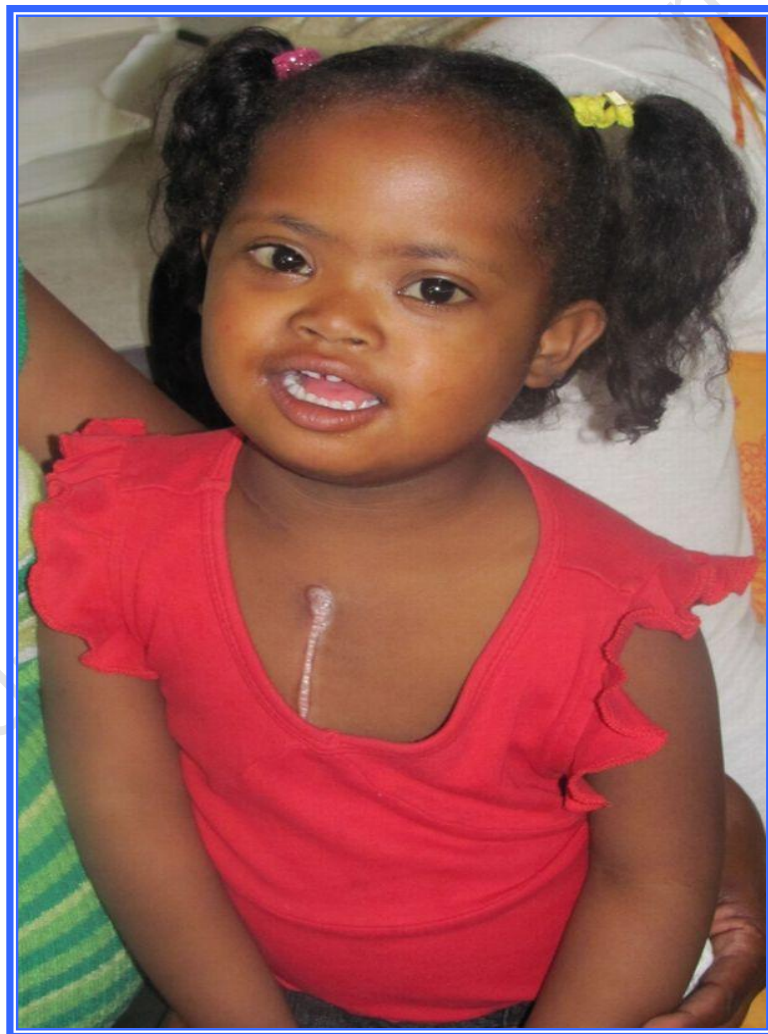
The research design entailed a phenomenological cross-sectional design using a multi-method approach of both quantitative and qualitative methods. Data was collected at one specific point in time by means of semi-structured interviews conducted with 12 mixed ancestry parents of a preschool child with DS. Convenience and purposeful sampling methods were used to recruit these participants from the population group who make use of the health care and supportive services at the RCWMCH and TBH. Each interview was audio-recorded and transcribed by the researcher. Content analysis was used to derive meaningful conclusions from the data.

#### **1.5 ORGANISATION OF THE STUDY**

In Chapter Two a literature review is presented on various aspects of DS. In Chapter Three the methodology of this study is described. The procedure of the entire research study is described, including the process of recruiting participants and a description of the measurement instruments in terms of appropriateness and validity/trustworthiness. Also provided is a brief explanation of the data collection and data analysis procedure. In Chapter Four the analysis and findings are presented and discussed. The main concluding findings are summarised in Chapter Five. Future recommendations as a result of the outcome of the study are discussed in Chapter Six.

## Chapter 2

# LITERATURE REVIEW

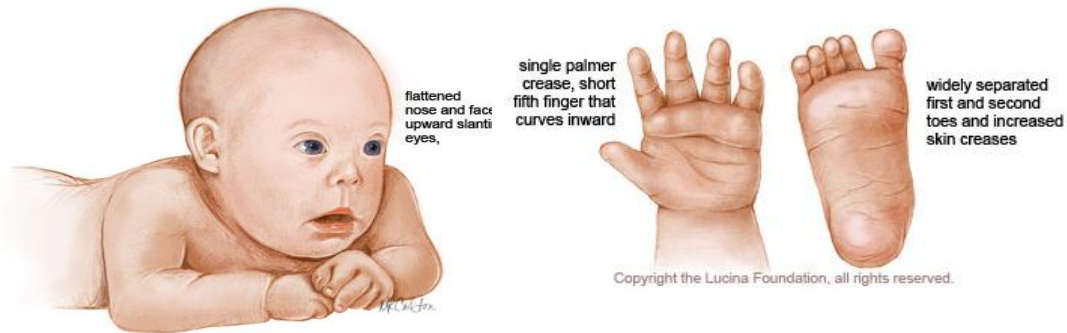


### 2.1 INTRODUCTION

This chapter includes literature reviews of the clinical aspects of DS, including the causes, diagnosis and management thereof. It also elaborates on the psychosocial impact of DS on the family, as well as the parents' satisfaction with the counselling and health care services. Preventive strategies are described including parents' perspectives regarding these options. Literature searches were performed on PubMed, Google Scholar, SA ePublications and ScienceDirect using "social support", "psychosocial impact", "Down Syndrome", "satisfaction with health care services", "satisfaction with delivery of diagnosis", "understanding of Down syndrome" and "attitudes towards prenatal screening and testing". All the way through this chapter the research that has been described was conducted in first world countries and not in SA, unless otherwise stated.

### 2.2 CLINICAL ASPECTS OF DS

The distinct clinical features of DS was first described in a group of children with an intellectual disability by Dr John Langdon Down (1866) and now bears his name (Chudley and Chodirker 2003). Their unique collection of features include: hypotonia, small brachycephalic head, a protruding tongue, hyperflexibility of joints, short stature and abnormal gait (Jones 1997). Individuals with DS have characteristic facial features such as epicanthal folds, upslanted palpebral fissures, flat nasal bridge and upper-mid face (Figure 2.1). Other distinct features include single palmer crease (Figure 2.1), clinodactyly, short fingers and thumbs, sandal gap (Figure 2.1) and excessive skin at the nape of the neck (Pueschel and Murphy 1976; Winship 2003).



**Figure 2.1: Characteristic facial and physical features seen in children with DS**  
 (<http://www.hindustanlink.com/health-care-blog>)

All individuals with DS have an intellectual disability as well as a developmental delay. The average age for independent sitting is 6-13 months, walking 1-4 years and first words 1-3 years (Firth et al 2007). The degree of intellectual impairment varies between individuals, ranging from mild to moderate and rarely to severe (Committee on Genetics, American Academy of Paediatrics (AAP) 2001). The mean IQ is 45-48, with a wide range and upper limit of approximately 70 (Firth et al 2007).

Behavioural problems have been described as being more prevalent in children with DS compared to typically-developing children (Courtenay et al 2009). These problems include disruptive behavioural disorders such as attention deficit hyperactivity disorder (25%), autism spectrum disorders (10%) and oppositional defiant disorder (Dykens 2007). The negative aspects of their behaviour have been described as attention-seeking, hyperactivity, disobedience, stubbornness, impulsivity, aggression and inattention (Cuskelly and Dadds 1992; Dykens et al 2002). The positive characteristics are usually given as good-natured, affectionate, outgoing, happy and sociable (Wishart and Johnson 1990; Myers and Pueschel 1991).

Individuals with DS can also present with a variety of medical conditions and congenital malformations (Johnson et al 2006). Multiple systems are affected, but the extent to which each system is involved is highly variable from person to person (Marder and Dennis 1997).

### a) Congenital Heart Defects

Fifty percent of babies with DS are born with a heart problem, of which half are serious and require surgery (Firth et al 2007). This is the most common cause of death during the first two years of life (Chen 2009). The most common heart defects and their respective frequencies are presented in Table 2.1 (Marder and Dennis 1997; Chen 2009).

**Table 2.1: Most common congenital heart defects (CHD) in individuals with DS (Chen 2009)**

CHD	Reported frequency
Endocardial cushion defect	43%
Ventricular septal defect	32%
Atrial septal defect	10%
Tetralogy of Fallot	6%
Isolated patent ductus arteriosus	4%

### b) Ear, Nose and Throat Problems

*Ears:* Approximately 75% of children with DS experience auditory problems of which chronic otitis media and hearing loss are the most common (Wexler et al 2009). Hearing loss is mostly acquired from recurrent infections, but a few cases of sensorineural deafness have been reported (Marder and Dennis 1997). Assessment by means of the auditory brainstem response (ABR) has indicated that 66%-89% of the children with DS have a hearing loss of greater than 15-20dB in at least one ear (Chen 2009).

*Upper airway obstruction:* Due to the narrow airway passages and tongue hypotonia in individuals with DS they are particularly prone to partial upper airway obstruction and 50-75% develops obstructive sleep apnoea (Stebbens et al 1991; Van Cleve and Cohen 2006). Growth retardation, poor developmental progress, tiredness and lethargy may occur, as well as significant hypoxia and cor pulmonale (Marder and Dennis 1997).



### c) Ophthalmological Problems

Ophthalmological problems are prevalent amongst 60% of individuals with DS (Wexler et al 2009). The most common eye problems and their frequencies are listed in Table 2.2 (Committee on Genetics, AAP 2001; Chen 2009). Other problems that are more prevalent in DS are conjunctivitis, blocked nasolacrimal ducts, glaucoma and acquired lens opacity (Marder and Dennis 1997).

**Table 2.2: Most common ophthalmologic problems seen in DS (Chen 2009)**

Ophthalmologic problem	Reported frequency
Refractive errors	50%
Strabismus	44%
Blepharitis	33%
Nystagmus	20%
Cataracts	15%

### d) Gastrointestinal Abnormalities

Twelve per cent of children with DS have congenital gastrointestinal malformations, including duodenal atresia or stenosis, imperforate anus, annular pancreas and exomphalos (Marder and Dennis 1997; Committee on Genetics, AAP 2001). These complications may be identified on antenatal scans or may present in the neonatal period. Many infants present with feeding difficulties due to oral motor dysfunction or gastro-oesophageal reflux. Also more common in DS are coeliac disease (~4%) and Hirschsprung's disease (~2%) (Marder and Dennis 1997).

### e) Other Medical Problems

Thyroid dysfunction, especially hypothyroidism occurs in 15% of individuals with DS (Stewart 1994; Wexler et al 2009). Obesity is observed in 60%-80% and constipation in 44% of individuals (Johnson et al 2006). The incidence of leukaemia is about 20 times higher than that of the general population and includes both acute lymphocytic

leukaemia (~2%) and acute non-lymphocytic leukaemia (Firth et al 2007). Six percent of children with DS present with acquired hip dislocation and infantile spasms occur in 10% (Committee on Genetics, AAP 2001; Chen 2009). Individuals with DS are also at an increased risk of developing infectious diseases, especially pneumonia (Chen 2009). Adults with DS are at an increased risk of developing dementia similar to that of Alzheimer disease (Firth et al 2007).

### **2.2.1 Diagnosis of DS**

DS is most often suspected at birth or in the newborn period (Marder and Dennis 1997). When considering a diagnosis of DS it is also important to take a detailed family and pregnancy history as well as documenting the mother's age at the time of the child's birth. This information is helpful when trying to establish the cause of DS (Firth et al 2007).

A confident clinical diagnosis is usually made after a thorough physical examination of the infant. The diagnosis is then confirmed by chromosomal analysis (Marder and Dennis 1997). This entails drawing blood from the child and examining the full karyotype under the microscope. If the blood karyotype is normal it is important to investigate the possibility of DS mosaicism by doing a skin biopsy for a fibroblast karyotype (Firth et al 2007).

In some cases DS may be suspected antenatally due to findings on ultrasound examination or maternal blood testing. In these cases a prenatal diagnosis can be made by obtaining foetal cells via amniocentesis, chorionic villus sampling (CVS) or cordocentesis techniques, culturing the cells and obtaining a karyotype from them (Norrgard 2008).

### **2.2.2 Management of DS**

DS is an incurable condition and therefore management mainly involves quality routine assessment and monitoring as well as ongoing medical management specifically tailored

to the needs of each individual (Van Cleve and Cohen 2006). A number of guidelines and protocols have been drawn up suggesting the appropriate medical surveillance and management aimed at identifying problems at an early stage so as to prevent the occurrence of secondary complications (Pueschel et al 1995; Howells 1996; Cohen 1999; Van Cleve and Cohen 2006). Table 2.3 is an example of these protocols (Marder and Dennis 1997).

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**Table 2.3: Example of a programme of care for children with DS in developed countries (Marder and Dennis 1997)**

Age	Topics for discussion	Examination	Tests	Referrals
<b>Neonatal</b> See in first few days of life Follow-up at 2-3weeks	Explanation of diagnosis Genetics Development Medical problems Local resources Contact with DSA	Routine neonatal examination: special attention to look for cardiac problems and cataracts	Chromosomes Thyroid function Neonatal hearing screen ECG, CXR, Echocardiogram	Paediatric cardiologist Medical geneticist DS service Social worker if necessary Notify primary health care team
<b>First Year</b> Review at 3 and 6 months	Parental concerns Feeding Hearing and vision Cardiac assessment results Family adjustment	Cardiac Eyes: visual behaviour, squint, nystagmus, cataract Growth Development	Hearing at 8 to 10 months	Genetic counselling Speech therapy Physiotherapy or occupational therapy as necessary Preschool education team
<b>Preschool</b> Annual medical review	Parental concerns Developmental progress General health, especially ENT Hearing and vision	Eyes Growth ENT	Hearing: annually Vision: annually from 2 years Thyroid function: 2 years	Ophthalmology: age 2 years Formal notification to school of choice
<b>School Age</b> Annual medical review	Parental concerns Developmental progress General health, especially ENT Hearing and vision Parental	Eyes Growth ENT Neurology: signs of compression	Hearing: 1-2yearly Vision: 1-2yearly Thyroid: 3yearly	Only as clinically indicated

From Table 2.3 it is clear that the effective management of children with DS involves a multidisciplinary team of health care professionals as well as a variety of supportive services (Marder and Dennis 1997). These children are introduced to the early intervention programmes run by various hospitals as well as the Down Syndrome Association (DSA). Occupational, physical and speech therapy as well as special education, nutritional and social work support are some of the specialised programmes and related resources available to children with DS as part of the early intervention programme (Van Cleve and Cohen 2006).

### **2.2.3 Life Expectancy and Prognosis of DS**

The life expectancy of people with DS has been increasing since the 1920s due to better healthcare in treating CHD and other physical health disorders and better education and provision of services (Coppus et al 2008).

In first world countries the median age of death is 49 years, with 44.4% surviving to the age of 60 years and 13.6% to the age of 68 years (Yang et al 2002; Firth et al 2007). Australian data have shown that 75% of individuals with DS survive to the age of 50 years (Glasson et al 2003). However in lower resource nations, the mortality in infancy and early childhood is far greater (Christianson and Modell 2004). In SA it has been reported that two out of three children with DS die before the age of two years (Christianson et al 2002).

Even though DS remains a lifelong chronic disability which reduces life expectancy, the overall outlook for these individuals with DS has dramatically improved (Wexler et al 2009). Individuals are generally healthier and better integrated into society (Chen 2009). There is also a greater recognition that with the appropriate early medical, rehabilitative and educational interventions, individuals with DS can lead relatively normal and productive lives (Wexler et al 2009). Most teenagers with DS achieve a degree of independence, learn to dress themselves and speak so that their family can understand them. However in adult life they require some form of supervision on a daily basis and some may be able to work in a sheltered environment (Firth et al 2007).

## **2.3 CAUSES OF DS**

In 1959, Lejeune et al (1959) first discovered that the cause of DS is trisomy of chromosome 21. This triplication can either be complete or partial, which results from one of three mechanisms: non-disjunction, translocation or mosaicism (Van Cleve and Cohen 2006; Bittles et al 2007). It is important to determine the cause of DS as it has implications for the recurrence risk of DS in the family.

### **2.3.1 Non-disjunction**

In 95% of individuals with DS their condition is caused by a complete extra chromosome 21 in all cells, due to a non-disjunction event during meiosis (Van Cleve and Cohen 2006). Most commonly the error is maternal non-disjunction during meiosis I and less frequently meiosis II. Few cases have been reported where the extra chromosome has resulted from paternal meiosis II errors (Chen 2009).

The recurrence risk of this type of DS is directly affected by maternal age. It has been reported that the incidence of DS caused by non-disjunction increases significantly with an increase in maternal age (Table 2.4). Women who have had a child with DS who are younger than 39 years of age are informed of a recurrence risk of slightly less than 1% and women who are older than 39 years are given an age-related risk based on the odds reported in Table 2.4 (Firth et al 2007).

**Table 2.4: Observed and predicted odds of DS live births by maternal age (Adapted from Morris et al 2002).**

Maternal age at child's birth (in years)	Observed odds	Predicted odds
20	1:1441	1:1476
30	1:959	1:937
35	1:338	1:352
36	1:259	1:266
37	1:201	1:199
38	1:162	1:148
39	1:113	1:111
40	1:84	1:85
41	1:69	1:67
42	1:52	1:54
43	1:37	1:45
44	1:38	1:39
45	1:32	1:35
46	1:31	1:31

### 2.3.2 Translocation

An unbalanced Robertsonian translocation involving chromosome 21 accounts for approximately 3%-4% of cases of DS (Committee on Genetics, AAP 2001). This occurs when genetic material from chromosome 21 attaches itself to another chromosome. Most commonly it attaches itself to another acrocentric chromosome, most frequently chromosome 14; less frequently chromosome 22, 13 or 15; and rarely the other chromosome 21 (Jones 1997). The great majority of these translocations are *de novo* and approximately 1 in 4 is the result of a familial translocation (Committee on Genetics, AAP 2001).

The chances of recurrence depend on the type of translocation, which chromosomes are involved and whether one of the parents carries the translocation. In the case of a *de novo* Robertsonian translocation involving 21, the recurrence risk is given to the parents as less than 1%. If it is found that the mother is a balanced carrier for the translocation involving 21, the recurrence risk is given as 10%-15%. If the father is identified as a

carrier, the recurrence risk is less than 1% (Firth et al 2007). There is a 100% recurrence risk if either parent is a balanced carrier for the Robertsonian 21:21 translocation (Harper 2004).

### **2.3.3 Mosaicism**

Mosaicism is the least common cause of DS and occurs in only about 1%-2% of individuals (Van Cleve and Cohen 2006). There are two different cell lines present in these individuals: the one normal and the other with trisomy 21 (Committee on Genetics, AAP 2001). In the majority of cases this is due to postzygotic non-disjunction or to a lesser extent, postzygotic loss of a chromosome 21 from a trisomic zygote (Firth et al 2007). The recurrence risk for mosaic DS is estimated to be 1% (Jones 1997).

## **2.4 PREVENTIVE STRATEGIES FOR DS**

The perception of DS as a public health concern has led to the development of a prenatal testing industry. The majority of countries have developed prenatal screening policies and equitable access to DS screening is often seen as a benchmark for quality antenatal services (Bryant et al 2006). There are two main aspects to these screening programmes: prenatal screening testing and prenatal diagnostic testing. The former refers to the probability that the foetus has DS and the latter to the diagnosis of DS in the foetus (Sooben 2010).

### **2.4.1 Prenatal Screening Testing**

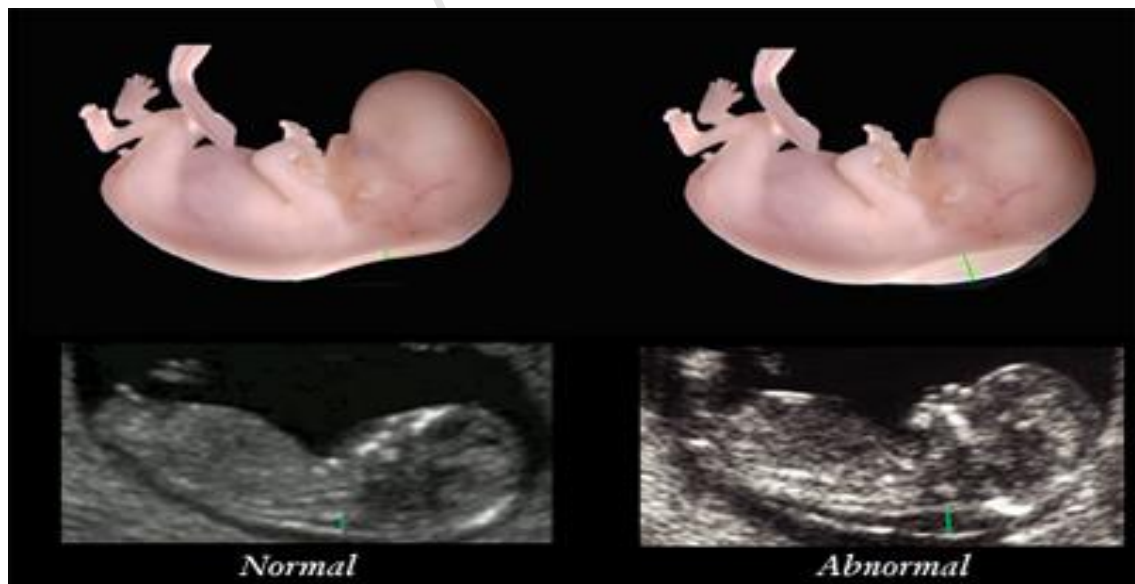
Prenatal screening options are offered to all pregnant women as part of their routine antenatal care. These screening programmes aim at identifying mothers at-risk of having a child with DS so that they may receive genetic counselling regarding their specific circumstances and discuss further available management options (Christianson and Modell 2004). The screening tests are offered according to the gestational period of the pregnancy and usually entail determination of advanced maternal age (AMA), biochemical screening and ultrasound scanning (Lampret 2006).



#### 2.4.1.1 First Trimester Screening

In SA the first trimester screening tests differ in the public and private health sectors. In the public sector, biochemical screening is not offered. In the private sector, first trimester biochemical screening for DS has been established by measuring the levels of pregnancy-associated plasma protein-A (PAPP-A) and the free beta subunit of human chorionic gonadotrophin (f $\beta$ -hCG) present in the maternal blood. It has been found that in the pregnancies with foetuses affected with DS, there are reduced levels of PAPP-A and increased levels of f $\beta$ -hCG present (Wheeler and Sinosich 1998).

An ultrasound marker that is being effectively used for screening during the first trimester is nuchal translucency (NT). The NT scan (Figure 2.2) can only be done by appropriately trained operators and is only offered between 11 and 14 weeks gestation. This scan involves measuring the thickness of a fluid filled space behind the neck of the foetus. An increase in fluid collection is associated with an increased risk for chromosomal anomalies, including DS, as well as congenital heart disease and skeletal dysplasias (Nicolaidis et al 1999).



**Figure 2.2: Nuchal Translucency scans indicating the difference in thickness between normal and abnormal collection of fluid.**

Studies have shown that 90% of foetuses with trisomy 21 and other major chromosomal abnormalities can be identified by using a combination of NT, PAPP-A and f $\beta$ -hCG, with a false-positive rate of 5% (Nicolaides 2005).

### ***2.4.1.2 Second Trimester Screening***

During the second trimester of pregnancy the maternal serum triple test is used to screen for DS. This test includes the measurement of AFP, human chorionic gonadotrophin (hCG) and estriol levels present in maternal blood during 16 and 18 weeks gestation. Low levels of AFP and estriol and high levels of hCG are indicative of a high risk for DS (Haddow et al 1992).

Between 18 and 20 weeks gestation, foetal anomaly scanning is performed. For the detection of soft markers indicative of DS it is important for these scans to be performed by skilled operators using high quality equipment (Lampret 2006). Approximately 50%-70% of cases of DS are diagnosed by detecting soft markers during second trimester ultrasound (Shipp and Benacerraf 2002).

### **2.4.2 Prenatal Diagnostic Testing**

Women who are at an increased risk of having a child with DS are offered a variety of prenatal diagnostic tests and if applicable termination of pregnancy (TOP). The invasive diagnostic techniques most often used include CVS, amniocentesis and cordocentesis (Wilson et al 2005). These testing methods enable the physicians to make a definite diagnosis as early as the first trimester of pregnancy (Hook et al 1983; Christianson 1996). Amniocentesis and CVS are the two most commonly used techniques to obtain foetal cells to determine the foetal karyotype for the prenatal diagnosis of DS and other chromosomal anomalies (Brock et al 1992; Harper 2004; Lo et al 2009). A short summary of each procedure is provided in Table 2.5, including the advantages, risks and limitations of each (Harper 2004; Wilson et al 2005).

**Table 2.5: Summary of Amniocentesis and CVS Information (Harper 2004; Wilson et al 2005)**

	<b>Amniocentesis</b>	<b>CVS</b>
<b>Procedure</b>	Ultrasound guided removal of amniotic fluid by needle and syringe	Ultrasound guided removal of chorionic villi by transcervical catheter (TC) or biopsy forceps (BF) and syringe or transabdominal (TA) needle insertion
<b>Timing (gestational period)</b>	15-20 weeks	TA: 10-32 weeks TC: 10-12 weeks
<b>Added risk of miscarriage due to procedure</b>	0.5%-1.0%	TA: 1%-2% TC: 2%-6%
<b>Foetal malformation risks</b>	—	1 in 3000 vascular limb malformation (suggested but not proven)
<b>Chance of successful sampling</b>	Approximately 99%	Approximately 99%. If unsuccessful, can follow with amniocentesis.
<b>Time required for cytogenetics diagnosis</b>	1-3 weeks (FISH available in days)	2-3 weeks
<b>Accuracy (chromosomes)</b>	Highly accurate: 95.5%	Highly accurate: 96%-98%
<b>Mosaicism</b>	True foetal mosaicism rare	Confined placental: 1%-2%
<b>Open neural tube defects (NTD's)</b>	AFP in amniotic fluid detects approximately 95% of NTD's	Other tests required for NTDs

Currently attempts are being made at developing Non-invasive prenatal diagnosis techniques for diagnosing aneuploidies, such as DS. These techniques involve collecting cell-free DNA from the foetus found in the bloodplasma of the pregnant woman (Lo et al 1997; Wright and Chitty 2009). The basis of these tests is the detection of foetal-specific DNA sequences in maternal plasma (Lo and Chiu 2007). The aim is detect the presence of an elevated amount of chromosome 21 sequences, as there is three rather than two copies of foetal chromosome 21 (Lo et al 2007). These techniques will not be discussed further as these testing options have not been successfully established as an appropriate diagnostic tool and is not available in SA (Chiu et al 2011).

### **2.5 PSYCHOSOCIAL IMPACT OF DS**

During a pregnancy most parents experience hopes and dreams for their unborn child. The birth of a baby with a disability, such as DS, tends to shatter any such dreams. In the hours after birth, as well as during the years ahead, parents and other family members not only have to learn to accept the child with DS, but also have to tolerate the social stigma and societal inequalities associated with the rights of people who are disabled (Ross and Deverell 2010).

#### **2.5.1 Impact on parents**

When a diagnosis is made of a disability in a child it usually destroys the parents' cherished dream of a healthy normal developing child. It is the loss of this dream that must be mourned by the parents. Grieving is essential for these parents so that they can separate themselves from the lost dream and thereby learn to accept the child as he or she is (Ross and Deverell 2010).

The main stages of the grieving process are described by Elizabeth Kubler-Ross (1969) as: denial, bargaining, guilt, depression, anger and acceptance. These different states are experienced in a cyclical fashion. Parents may oscillate between these emotional stages and a specific event in their child's life can once again force them into an emotional state that they have already successfully negotiated in the past (Ross and Deverell 2010).

Initially parents experience a period of grief and mourning and often feel there is no way out of the dilemma. This reaction of shock and numbness is believed to be a mechanism that protects the individual from experiencing further pain that he or she is unable to manage at that particular time (Steenkamp and Steenkamp 1992). Often these initial feelings are accompanied by tremendous sorrow and weeping (Ross and Deverell 2010). It is important for most parents to go through this initial phase of grieving to facilitate the process of acceptance of the child (Ross and Deverell 2010).

Parents do not only experience intense feelings of loss, but some may start blaming themselves or others for their child's condition and they experience strong feelings of guilt (Steenkamp and Steenkamp 1992). These feelings of guilt may lead to the belief that their child has DS as a form of divine punishment for sins that were previously committed (Ross and Deverell 2010).

Other parents may experience anger and embitterment, because they have a child with DS. They feel frustrated that a disabled child has uninvitedly entered the home and disrupted a once happy household (Steenkamp and Steenkamp 1992). Some parents may feel ashamed to be seen in public with their child because of societal perceptions of DS and these children being unwelcome (Sari et al 2006).

Many parents exhibit signs of denial of their child's diagnosis and try to convince their friends and the health care professionals that their child is no different from the other children in their neighbourhood. Denial can be detrimental to the child's health as it may prevent parents from making rational decisions about their child's management and future (Ross and Deverell 2010).

Some parents with children with DS have also been described as being in a state of prolonged crisis which alters and lowers their self-esteem, leads to depression and social isolation (Van Riper 2007).

Parents initially experience the above mentioned negative outcomes, but these adjustment problems are expected to decrease in time and positive outcomes, such as psychological well-being, personal growth, improved relations with others, changes in philosophical or spiritual views and satisfaction with parenting are likely to be experienced (Van Riper 2007). The strengths of parents of children with an intellectual disability, including DS, have been described as experiencing feelings of joy, hope, happiness and optimism as well as having more patience, a greater appreciation for the simple things in life and increased compassion (Abott and Meredith 1986; Van Riper 2007).

Some parents may only experience a few of the emotional reactions described, while others may be confronted by all of them. It is important to bear in mind that all these reactions are normal and acceptable (Ross and Deverell 2010). With time, parents generally learn to accept and cope with their child with DS and may even demonstrate attachment to their child (Roizen and Patterson 2003).

### **2.5.2 Impact on Marital Relationship**

Studies have indicated that there is no difference in the marital functioning of couples with a child with DS compared to that of the general population (Van Riper et al 1992; Ross and Deverell 2010). Kersh et al (2006) also indicated that there is no difference in marital functioning, but that there is a lower level of marital quality than in the general population.

However there have been a small number of separations seen to occur, usually due to neglect of a partner, due to the excessive devotion of the other partner to the child with DS (Sari et al 2006). The other problems that married couples experience are usually related to problems that were present before the birth of the child with DS. These factors are aggravated by the stress of having a child with DS and often lead to increased marital dysfunction (Ross and Deverell 2010).

### **2.5.3 Impact on Siblings**

Siblings of children with a disability are believed to react in one of three ways. The first being resentment due to the lack of attention given to them by their parents which, in turn, may lead to behavioural problems, attention-seeking behaviour or withdrawal from the family due to feelings of rejection (Sari et al 2006). The second reaction is one of shame and embarrassment to be seen with their disabled sibling. The third reaction is where the healthy sibling will imitate the disabled sibling in an attempt to get more attention (Ross and Deverell 2010). Fisman (1996) described those siblings who believed that their sibling with DS is preferred by their parents, as having more anxieties and a lower self-esteem than the siblings in the control group.

Skotko and Levine (2006) found that siblings of persons with DS in Boston experienced both positive and negative feelings with their sibling relationships, but the positive emotions most often outweighed the negative ones. A study done in Australia by Cuskelly and Gunn (2003) supported this view and indicated that siblings of a child with DS exhibited more kindness, empathy and caregiving activities towards their sibling than controls. Siblings have favourable self concepts and many believe they develop additional strengths because of their sibling with DS (Erikson and Upshur 1989; Cuskelly and Dadds 1992; Van Riper et al 1992; Skotko and Levine 2006).

### **2.5.4 Impact on Family Functioning**

The family systems theory states that any disorder affecting one subunit of the family compromises all other units in the system (Sari et al 2006). In the case of DS where the individual's physical and cognitive constraints limit him or her from fully engaging in family, social and community interactions, so too do many of the other family members experience significant alterations in their daily lives (Marder and Cholmain 2006). The presence of a child with DS may therefore lead to major social, economic and emotional difficulties, and hence a remarkable change in the functioning of the family as a whole (Sari et al 2006).

Cuskelly et al (2008) has identified several important domains of family life: health, financial well-being, family relationships, religious and cultural beliefs, social support, leisure enjoyment, and community involvement. Brown et al (2006) reported that families of children with DS have a lower quality of life in relation to all of these domains compared to a comparison group of families with normal developing children.

Economic resources are known to contribute to the well-being of families. Of these, employment is one of the major resources as it connects the individual to the outside community (Cuskelly et al 2008). Warfield (2001) reported that because having a child with DS puts greater demands on the parents it leads to greater absenteeism from work. They also found that only two-thirds of the parents were employed full-time. An American study done by Schieve et al (2010) showed that 40% of the parents stopped working because of their child's condition and, in over one third; the family member had to reduce their working hours. Overall 40% of these families reported family financial problems caused by their child with a disability.

On account of the strong social stigma that is attached to intellectual disability, some families may feel ashamed to be seen in public with their child with DS and may become isolated from the community. However this reaction is usually not only due to the direct rejection by family and friends, it may be due to the parents' own fear of rejection and desire to isolate themselves and place a protective barrier around their child (Sari et al 2006).

Family life in families of children with DS is likely to contain some mix of hassles and uplifts, disappointments and great satisfactions, and it is therefore important that the uplifts and satisfactions be considered if an accurate picture of family life is to be gained (Cuskelly et al 2008). A study done by Cunningham (1996) found that the majority of families with a child with DS did not exhibit pathological function and evidence pointed to the member with DS having positive effects on the family. Van Riper (1999) described having a child with DS as a positive and growth-producing experience for



families. Poehlmann et al (2005) reported that mothers felt that their child with DS acted to maintain and develop connections between family members and others.

### **2.5.5 Importance of Social Support**

Social support is defined as the information leading the person to believe that he is cared for, loved, valued, esteemed and is important in a network of mutual obligation and communication (Cobb 1976). Examples of social support include: support from one's partner, extended family, and friends, and the availability of time to participate in recreational activities as well as support from community programmes or parent organisations, religious groups, health care professionals and the availability of services and programmes specifically aimed to help families of children with DS (Siklos and Kerns 2006; Ross and Deverell 2010). Agosta (1983) identified three fundamentals that social and family supportive services should embody: (1) services should both enable and empower parents to make informed decisions regarding their disabled child; (2) services should be open to addressing the needs of the entire family; and (3) services should be flexible in accommodating the unique needs of the family and disabled child.

Research has suggested that the range of available emotional and practical supports outside of the family significantly influence family well-being (Buckley 2002). Evidence has showed that support by friends, relatives and health care professionals play an important role in assisting mothers in their role as parents of a child with a disability and that successful grieving depends on the amount of human interaction (Skotko 2005, Ross and Deverell 2010).

Support provided by the extended family of grandparents, aunts and uncles can be a very positive source of support in the majority of families. Equally so it can also be a major stressor in some families if the grandparents or other members cannot accept the baby and are pessimistic about the future. The family's existing networks of friends in the neighbourhood, at church, play schools, mother and toddler groups and at clubs in the community can also be a very positive source of support provided that they are positive about the child with DS. Another important source of support for families is support

groups or DS associations. Some parents have reported that the most significant emotional and practical support that they had received came from other parents with older or similar aged children with DS sharing practical tips and advice (Buckley 2002).

Unfortunately previous studies have found that many parents feel that the available social services do not adequately address their needs. Freedman and Boyer (2000) reported that parents felt their needs were being unmet in the areas of types of services and support, socialisation and community inclusion opportunities for their child. A study done by Siklos and Kerns (2006) indicated that families of children with developmental disabilities felt that the service delivery systems did not provide them with adequate financial and social support.

### **2.6 GENETIC COUNSELLING**

According to the National Society of Genetic Counselors (NSGC) genetic counselling can be defined as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence;
- Education about inheritance, testing, management, prevention, resources and research; and
- Counselling to promote informed choices and adaptation to the risk or condition.”

(Resta et al 2006:77)

Genetic counselling is an important part of managing families of children with DS (McGrath et al 2009). The counselling is done by various health care professionals including paediatricians, clinical geneticists, obstetricians and genetic counsellors (Committee on Genetics, APA 2001). The following topics should be discussed with the families:

- Diagnosis of DS and chromosomal karyotype results;

- Mechanism for occurrence of the disorder in the child and the potential recurrence risk for the family;
- Manifestations of DS and associated medical conditions, including variability seen amongst infants, as well as prognosis;
- Currently available treatments and interventions, including the effectiveness, complications, adverse effects and costs of each;
- Early intervention programmes and available parent support groups; and
- The options available to the family for management and rearing of the child using a nondirective approach. (Committee on Genetics, APA 2001; Van Cleve and Cohen 2006).

### **2.7 LEVEL OF UNDERSTANDING OF GENETICS**

Genetic concepts in general are complicated and difficult to understand and most members of the public do not even know basic concepts of genetics (for example that genes reside in every cell of the body) and that humans have 46 chromosomes (Kessler et al 2007).

Parents of a child with a birth defect, such as DS, are likely to have many questions about the genetic cause, prognosis and recurrence risks. The genetic counselling service is aimed at helping these parents understand difficult genetic concepts surrounding their child's condition as well as answering some of their questions and allowing parental concerns and anxieties to be addressed (Harper 2004).

Several studies have been undertaken to evaluate the level of understanding of parents after the initial genetic counselling session. The majority of the studies have indicated that the researchers consider this level is unsatisfactory and have identified various factors that influence parents' understanding of the information given (Oetting and Steele 1982; De Pina-Neto and Petean 1999; Molster et al. 2009; Klitzman 2010).

Oetting and Steele (1982) investigated the level of general genetic knowledge and DS amongst counselled and non-counselled couples. They found that there was no significant difference between the knowledge of general genetics and the knowledge of recurrence risks of DS between the counselled and non-counselled groups. They did however show that the ability of the two groups to recall the information was directly linked to the time elapsed since their initial genetic counselling session. De Pina-Neto and Petean (1999) indicated similar findings and showed that parents' knowledge of the genetic condition decreased over time.

Collins et al (2001) highlighted the importance of timing the genetic counselling session correctly. It is important to acknowledge that parents' ability to take in the information accurately will largely be influenced by their stage in the adaptation process. During the initial phases where feelings of shock, anger and denial are the greatest, a limited amount of information will be gained (Miller et al 1994). Klitzman (2010) also showed that the emotional state of the parents play an important role in their ability to grasp the information.

Lampret and Christianson (2007) investigated the level of knowledge of DS amongst SA mothers with a child with DS. They found that in most cases the mothers only had basic knowledge of DS. They found that ethnicity played a role and that 90% of the caucasian mothers and only 19% of the black women had known about DS before the birth of their child. Ethnicity and level of education is closely associated with each other within the context of SA. Black individuals have a lower level of education than Caucasian individuals (Lehohle 2004).

A study done by Molster et al (2009) in Australia found that the level of education of the participants as well as their level of income played a role in their understanding of genetic information. Higher income and educational levels lead to higher levels of understanding. Klitzman (2010) supported this view. De Pina-Neto and Petean (1999) had similar findings and concluded that a low socio-economic level was a major factor in reduced understanding.

Other factors believed to have an impact on the level of understanding are socially and culturally widespread beliefs as well as religious beliefs of the parents. Certain concepts and misunderstandings of genetic disease are promoted by parent support groups, the media, rumours and hearsay. Parents may also present with misconceptions regarding the causes of genetic conditions due to their religious beliefs and may think that the condition is a form of divine punishment (Klitzman 2010).

Parents' personal experiences and interactions with individuals with a specific genetic condition play a role in shaping their understanding. Lucke et al (2004) and Walter et al (2004) found that if parents' had more interaction with a person with a specific condition it increased their understanding thereof.

### **2.8 SATISFACTION WITH GENETIC COUNSELLING SERVICES**

Several studies have investigated the satisfaction parents experience with genetic counselling. Overall high levels of satisfaction with these services have been found (Bleiker et al 1997; Michie et al 1997; Stadler and Mulvihill 1998).

The effectiveness of genetic counselling in general was investigated by Davey et al (2005) by measuring the level of client satisfaction. They found that the majority of the clients were extremely satisfied with their overall experience. The overall satisfaction for most of these participants was determined by the degree of dedication by the counsellor, the interest of the counsellor in their personal problems and the way in which the information was communicated to them.

A study done amongst Australian parents of children with DS and Cystic Fibrosis (CF) indicated high levels of satisfaction with the counselling services (Collins et al 2001). They found that all of these parents experienced it positively and indicated that getting the information they needed to deal with the diagnosis and relief from guilt feelings were the most favourable aspects.

Collins et al (2001) found that the only complaints some parents had regarding the genetic counselling session was that at times too much information was given at once. Some parents felt that it would have been useful to have a genetic counselling session at a later stage when they were more at ease with their child with DS, since their children were very sick in their first year of life.

### **2.9 PARENTS' SATISFACTION WITH THE MANNER IN WHICH THE NEWS WAS BROKEN**

The way in which the news is delivered to the parents that their child has DS is an extremely important event in these parents' lives as they have a tendency to remember exactly how they were first told, the way this news was framed and the emotions they experienced (Muggli et al 2009).

The majority of families who have children with DS only learn of their diagnosis after birth despite prenatal diagnosis options being readily available. Delivering and receiving a diagnosis of DS is not an easy experience for either physician or the parents. For these parents the announcement is at minimum surprising and unexpected (Skotko 2005). Several studies have indicated that many parents report feeling shocked, anxious, guilty, frightened, confused, devastated and angry after learning of their child's diagnosis (Noble 1993; Burke and McDaniel 2001; Skotko 2005).

Extensive research has been done on the parents' experiences of this event and despite many protocols being written on how to improve this it is still found to be inadequate (Berg et al 1969; Pueschel and Murphy 1976; Krahn et al 1993; Garwick et al 1995; Van Riper 2003).

Pueschel and Murphy (1976) reported that out of 414 American mothers, approximately 40% felt the news was broken in an inadequate and unsympathetic way by their physicians. Murdoch (1984) reported similar findings in a survey of 123 Scottish mothers.

Skotko and Canal (2004) found that the majority of mothers felt frustrated and dissatisfied with their physician and felt that they weren't provided with enough up-to-date information after being told about their child's diagnosis. The researchers also found the mothers feeling more optimistic if their doctors emphasised the positive aspects of DS rather than the negative aspects.

Hedov (2002) and Muggli et al (2009) found that Swedish and Australian parents respectively were unhappy if the news was provided immediately after the baby's delivery before the mother's needs were seen to and she was still in pain from the labour. They were also dissatisfied if they were told before holding the baby for the first time. Muggli et al (2009) reported that all 18 Australian parents responded favourably to and remembered health professionals who sat down by their bed, listened to them, or made a special effort to follow-up their questions.

Parents have expressed intense dissatisfaction when they received the diagnosis in a public ward with other patients in the room as well as visitors (Garwick et al 1995; Skotko 2005). Mothers desired hearing the diagnosis in a room secluded from others and desired the company of their husband or partner when the news is broken (Cunningham et al 1984; Garwick et al 1995; Skotko et al 2009).

Other factors that contributed to how the parents experienced this event have been described and include level of education, socio-economic status, religion and whether the diagnosis was made prenatally or postnatally. It has been reported that the highly educated and wealthy American mothers, especially those in demanding careers, seemed to be more worried than the mothers who were less educated and wealthy. It has also been suggested that religion might impact on how a mother is able to cope with the new diagnosis as research has shown that Catholic and Christian mothers seem to cope better with the news than the Jewish, Protestant and Atheist mothers. Mothers also seemed happier over the birth of their infant with DS if they received a prenatal diagnosis than those who received the diagnosis postnatally as they tended to resolve any grief before the birth of their child (Skotko 2005).

### **2.10 PARENTS' SATISFACTION WITH HEALTH CARE SERVICES**

To promote parents' overall well-being it is important for them to feel satisfied with the health care services offered to them and their child with DS, as well as the ease of using these services (Pascoe 1983). Parents cope better if they feel they are receiving support from their health care providers and particularly value services that provide them with practical and realistic strategies for dealing with difficulties (Buckley 2002).

Unfortunately studies have indicated that a large number of parents with children with DS were unsatisfied with the health care services they received and that dealing with professionals had been a major source of additional stress (Stallard and Lenton 1992; Eaves et al 1996; Buckley 2002). The attitude of the professional, his or her way of working or the parent having to fight to get the service they want for their child all lead to increased parental stress (Buckley 2002).

Stallard and Lenton (1992) reported that parents felt they were receiving insufficient support, because the professionals did not have an understanding of the practical difficulties they face in their daily lives. Parents often felt that appropriate health care was not provided because the practitioner attributed the child's problems to the diagnosis of DS rather than to the actual health problem (Mason and Scior 2004). Wexler et al (2009) reported similar findings amongst 150 caregivers in Israel. They indicated that the caregivers felt that many of their children were not receiving appropriate medical follow-up and that the majority of health care professionals had a negative attitude towards DS.

Minnes and Steiner (2008) found that physician availability, knowledge of DS and services as well as the amount of time spent with the patient all affected the quality of care. They found that parents reported the experience more positively if the physicians were patient, accessible, willing to learn about their child's disability and able to make time to listen to caregivers. Eaves et al (1996) and Yam et al (2005) supported this and



found that the majority of parents in British Columbia and Hong Kong respectively were satisfied with the health care services offered to them and their child with DS.

### **2.11 ATTITUDES TOWARDS PRENATAL TESTING**

Various studies have focused on the views of the general public or pregnant women in general regarding prenatal testing options and the termination of an affected foetus. Very few of these studies specifically focused on the views of parents of a child with DS.

Julian-Reynier et al (1993) investigated the views amongst French women and found that those who had had a personal acquaintance or relationship with a child with a disability were less likely to undergo prenatal diagnostic testing. The relationship between parents and their child with a disability made testing more difficult for them, especially if they had a good relationship. Lo et al (2009) reported similar findings amongst pregnant Chinese women.

Several reports have been made that the attitudes of individuals towards prenatal testing are influenced by the parents' educational level, socio-economic status, ethnicity, previous obstetric history and age (Lampret and Christianson 2007; Lo et al 2009; Pieters et al 2009). Individuals of a lower educational level were less likely to opt for invasive diagnostic prenatal testing (Pieters et al 2009). Individuals of a higher educational and higher financial group expressed more positive attitudes towards testing (Lo et al 2009; Pruksanusak 2009). It has also been found that women who are younger were more positive than women who were older and of AMA (Pruksanusak 2009). Julian-Reynier et al (1994) found that women of a higher socio-economic status were more likely to accept invasive diagnostic testing than other women.

Lampret and Christianson (2007) investigated the views of 50 SA mothers living in Gauteng with a child with DS regarding prenatal diagnostic testing and the termination of a future affected pregnancy. They found that overall the majority (76%) of them would have prenatal diagnostic testing done. There were slight differences in views regarding TOP amongst the different ethnic groups, but the majority felt they would not

consider TOP. In the caucasian population group 52% would not consider TOP, in the black and Asian women 53% and 75% would not consider TOP respectively. These findings suggest that even though women would consider prenatal testing they may not have an intent to terminate an affected pregnancy. This was also found to be the case in a study conducted by Skotko (2005).

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## Chapter 3

# METHODOLOGY



### **3.1 INTRODUCTION**

In this chapter a description of the methodological process used to conduct this research study is provided. The reasons for having selected a specific method are provided in the relevant sections as well as the identification of potential sources of bias and attempts at minimising these.

### **3.2 RESEARCH DESIGN**

This research project was a phenomenological cross-sectional design using a multi-method approach of both quantitative and qualitative methods.

#### **3.4.1 Qualitative Approach**

A qualitative phenomenological approach was selected for this study as it aims to explore and describe people's understanding of experiences and behaviour, and the meanings and interpretations that they attach to these (Holloway 2008). This type of research permits the researcher to explore issues of interest in depth and detail without being constrained by predetermined categories and variables (Patton 1990). It allows the researcher to understand the richness and complexity of real-life events that occur in natural settings from the participants' points of view (McMillan and Schumacher 2001).

In this cross-sectional study data were collected at one specific point in time by means of interviews with the participants. It differs from longitudinal studies as data are not obtained from the same participants at several points in time (Bailey 1994). This method is preferred due to its simplicity and cost-effectiveness, but the disadvantage is that the change in the participants' social circumstances over time are not documented (Neuman 1999).

This research project was, according to published papers, the first of its kind in SA and therefore a qualitative method was selected. This approach is known to be especially valuable when first investigating a topic. Phenomenological research entails the

description of attitudes, emotions, perceptions, behaviours or other characteristics of a group of individuals regarding a current or past phenomenon (McMillan and Schumacher 2001).

### **3.3 SAMPLE**

#### **3.3.1 Population**

There are more than 100 children with DS attending the genetic clinics and supportive services at the RCWMCH and TBH. The majority of these children are of mixed ancestry or black ethnicity and their ages range from a few weeks to eight years of age.

Parents of 12 mixed ancestry preschool children with DS, who met the inclusion criteria, were identified by the allied health care professionals running the toy libraries at the respective hospitals.

#### **3.3.2 Eligibility Criteria**

##### *i) Inclusion criteria*

- Parents of children with DS between the ages of one and six years. This age range was selected because children within this age range need maximum intervention. Parents with children older than one year of age were selected so that they had time to utilise the health care services.
- Parents who had attended, or who were still attending, the health care and supportive services at the RCWMCH or TBH.
- Parents in the Cape Town metropolitan area of mixed ancestry ethnic backgrounds. This specific ethnic group was selected as it is representative of the vast majority of the parents attending the health care and supportive services and because experiences of having a child with DS in this ethnic group are not well-understood. In addition a specific ethnic group was selected, because ethnicity might have had an influence on parents' experiences.
- Parents who agreed to participate and consented to audio-recorded interviews.

- Parents with either Afrikaans or English as their home language, as the researcher is fluent in both.

*ii) Exclusion criteria*

- Parents of children with DS who were already attending school.
- Parents who had children with other medical conditions not generally associated with DS.

### **3.3.3 Sampling Method**

The sampling methods that were used included both purposeful and convenience sampling. Purposeful sampling aims at identifying information-rich cases who provide a full and sophisticated understanding of the phenomenon under study (Hansen 2006). This method allowed the selection of a small sample of participants to provide the best information available to address the specific purpose of this study. Participants were selected on the basis of being representative of the mixed ancestry population of preschool children with DS who made use of the health care and supportive services offered at the RCWMCH and TBH. They were also required to be accessible by attendance at support groups for the interviews. This method is known as convenience sampling (McMillan and Schumacher 2001).

The allied health care professionals providing the supportive services (Toy Libraries) helped identify the most suitable participants for this study during the period of July to October 2010. These participants were informed of the aims, objectives and method of the study by the health care professional running the Toy Library. The researcher then contacted the individuals who were willing to participate and re-explained the aims, objectives and method of the study. The researcher then contacted the first 12 individuals who were willing to participate to arrange a suitable date, time and venue for the interviews which took place during the period of September – November 2010. Seven of these participants were recruited from the services at the RCWMCH and five

from TBH. More than 12 parents met the inclusion criteria, but due to the time constraints of the study, only 12 could be included.

### **3.4 STUDY METHODS AND MEASURING INSTRUMENTS**

#### **3.4.1 Interviews**

The data were collected by means of a semi-structured interview schedule (Appendix III) adapted from interview schedules used in previous studies by the researcher. In qualitative research the one-to-one interview is often referred to as a conversation with a purpose. These interviews reflect the agenda of the researcher while obtaining in-depth data of the perspectives of the participants with regards to their life experiences of raising and living with a child with DS (Holloway 2008). Semi-structured interviews are a guide to help researchers remember topics they want to discuss during the interviews. This type of interview allows the researcher the freedom to ask additional questions, respond to issues or questions raised by the participants and to guide the conversation (Hansen 2006). The interview schedule combined both closed-ended and open-ended questions. Closed-ended questions were questions in which the responses were usually “yes” or “no” without encouraging elaboration (Polgar and Thomas 1991). Closed-ended questions were also asked to gather their sociodemographic details. Open-ended questions encouraged extensive responses without the limitation of preset answers and allowed parents to freely express themselves and elaborate on their experiences (Hansen 2006). These questions also provided the researcher the opportunity for in-depth questioning of the topic concerned. The discussion was guided by using prompt questions to gather the maximum amount of information in the allotted time (Smith et al 1995).

The interview schedule was adapted from interviews used in previous research studies done by Loggenberg (2006) and Schoeman (2007). The interviews used in previous research were used to investigate the experiences and knowledge of genetic information of parents of children with Duchenne Muscular Dystrophy and CF, respectively. It would not have been appropriate to use as is in the current research focusing on DS and

therefore had to be adapted to make it more appropriate to address the aim and objectives for this study. Particular questions from section D and E regarding the level of satisfaction of the health care services and social support respectively were adapted from a questionnaire used by Eaves et al (1996). The wording of these questions was changed to make it more appropriate for use in a semi-structured interview instead of a questionnaire with predetermined answers. In section G a measuring instrument was used to measure participants' general knowledge of DS. The participant was given a score for each specific characteristic of DS that they could correctly recall. The maximum scores for each subsection are indicated in Appendix III.

Content validity of the interview was established by the content being critically reviewed by the researcher's supervisors to ensure that the interview schedule was comprehensive and that all the questions were relevant and in an appropriate sequence (McDowell and Newell 1996).

All interview schedules (Appendix III), consent forms and information sheets (Appendix I) were available in basic English and Afrikaans to ensure that there was no ambiguity and they were easily understood by the participants. As the researcher is fluent in English and Afrikaans, the interviews were conducted in the language of the participant's choice.

### **3.4.2 Research Setting**

The interviews were conducted at a private venue of the participants' choice. Seven participants were interviewed in a private room at the RCWMCH at a time that coincided with a routine clinical visit. The other five participants were interviewed at the Down Syndrome Association Western Cape's offices at a time most convenient to them. These venues were found to be preferred by the participants as they found them more accessible and the environment less distracting than the home environment. The home environment would have been the researcher's preferred site as individuals are likely to feel more comfortable in this environment when responding to questions regarding



sensitive topics. A period of observation in this setting might also have given more information than what was obtained in the formal clinical setting (Smith et al 1995).

### **3.5 PROCEDURE**

The allied health care professionals at the respective toy libraries at the RCWMCH and TBH and the researcher informed the parents of the purpose and method of the study at a routine visit. The individuals willing to participate were then contacted by the researcher to arrange interview times and venues.

#### **3.5.1 Pilot Interviews**

A pilot study of two participants was conducted to test whether the items on the interview schedule were easily understood. While conducting these interviews the researcher took note of any signs such as body language and facial expressions indicating that they were uncomfortable or confused by the questions (McMillan and Schumacher 2001). Following the pilot study the interview schedules were adapted accordingly and certain questions were simplified to aid the understanding of the participants. The pilot interviews also ensured that no ambiguous questions were asked and helped to determine how much time was needed to complete an interview. In this study the interviews were found to take approximately 30-50 minutes. The data obtained from the pilot interviews were not included in the final results of this study.

#### **3.5.2 Recruitment**

The participants were recruited by the allied health care professionals involved in the Toy Library clinics at the RCWMCH and TBH, who had briefly explained the aims and objectives of the research study to them. The researcher attended these clinics during the recruitment period between July and October 2010, to meet with the recruited individuals and re-explain the aims and objectives of the research project. If both parents were available they were interviewed together. One parent was interviewed if only one

parent was available or in the case of single parents. The researcher also informed the parents that no extended family members should be present at the time of the interview.

### 3.5.3 Data Collection

Before conducting the interviews, written informed consent was obtained from each participant, which included permission to audio-record the conversations (Appendix I). At the beginning of each interview the participants were reassured that:

- The information provided during the audio-recorded interviews would be kept confidential apart from a possible publication in a scientific journal where no names would be used;
- The information would not be discussed with extended family members; and
- Participation was completely voluntary and that they were free to decide not to participate or withdraw from the study at any stage without it negatively affecting their access to the medical and supportive services to which they were entitled.

All the semi-structured interviews were personally conducted by the researcher. The interviews were audio-recorded as it provided a more complete record of the participants' exact words than hand written notes taken by the researcher (Smith et al 1995). It also enabled the researcher to maintain eye contact and concentrate on the participants' reactions (Holloway and Wheeler 1996). The interviews were transcribed and translated for analysis by the researcher. Quotations were "tidied up" and edited slightly so that the written format would be understandable to the reader (Denscombe 2008).

Due to the sensitive nature of some of the questions and the possibility of evoking anxiety, a follow-up visit was scheduled if the participants felt it was needed. The researcher contacted the participants one week to two weeks after their initial interview to clarify any uncertainties or concerns and to arrange a follow-up visit where needed.

The follow-up visit provided the opportunity for counselling the parents regarding any questions or emotions evoked during the initial interview.

### **3.6 DATA ANALYSIS**

Data analysis is the process by which the obtained data were transformed into meaningful new understandings, theories and statements about the empirical world (Hansen 2006). The transcripts itself were not meaningful as they only provided the raw data and a descriptive record of the research. For this data to become meaningful the researcher had to systematically sift through the transcripts of the interviews, interpret and capture the participants' responses (Pope et al 2000).

Content analysis was used to interpret the open-ended items data of the interview schedule, because it captured the richness of themes emerging from the interviews without reducing their responses to quantitative categories (Smith et al 1995). Analysis of closed-ended questions involved the selection of categories directly linked to the objectives of the study, and counting the frequency of these categories in the data (Hansen 2006). The data of this study were read and reread and index themes and categories were identified, which were centred on particular phrases, incidents or types of behaviour (Pope et al 2000). Inductive reasoning processes were used to interpret and derive meanings from the data (Thorne 2000).

In qualitative research a useful summary can be provided of some of the aspects of the analysis by using simple counts (Pope et al 2000). The responses to specific sections of the interview schedule were characterised by frequency and descriptive statistics such as percentages, means and averages to describe the occurrence of data.

### **3.7 TRUSTWORTHINESS/ VALIDITY**

In qualitative research trustworthiness reflects the measurement of its validity which refers to the truth, value and authenticity of the study (Holloway 1997). Validity refers to the degree to which the explanations of phenomena match the realities of the world

(McMillan and Schumacher 2001). Validity is also reflected by the extent to which the findings of the study are true to its aims and that they accurately reflect the purpose of the study (Green and Thorogood 2004).

The foundation for demonstrating trustworthiness in qualitative research is embodied in the following five elements: credibility, transferability, dependability, confirmability and authenticity (Lincoln and Guba 1985).

Credibility refers to the accuracy of the researcher's identification and description of the participants (Holloway 2008). In the current study this was ensured by peer debriefing and meetings with the supervisors on a regular basis to ensure the accurate interpretation of data.

Transferability means that the findings in this context can be applied to the experiences of other individuals in similar contexts and settings (Holloway 2008). By describing the data accurately and in extensive detail, peers and readers are provided with a full insight and a clear picture of how this research was conducted. This allows them to decide whether the findings may be transferred to other individuals.

The dependability of a study refers to the consistency and accuracy of the data over time. This was demonstrated by providing detailed descriptions of the methodology and thought processes involved (Holloway 1997).

Confirmability means that the researcher has represented the reality of the participants and that the findings are due to the results and that they were not influenced by the biases and subjectivity of the researcher (Holloway 1997; Holloway 2008). Transcriptions of audio-recordings and data analysis were confirmed by the researcher's supervisors.

Authenticity is established when the researcher reports the participants' perspectives fairly and when it helps them to understand and improve their condition by empowering

them (Lincoln and Guba 1985). This was obtained by follow-up telephone calls and by presenting the findings to the participants to confirm that they agreed with the researcher's interpretation of what was said during the interviews.

### **3.8 ETHICAL CONSIDERATIONS**

#### **3.8.1 Ethical Approval**

This study was granted approval without reservations by the Research Committee of the Department of Clinical Laboratory Sciences and the Research and Ethics Committee of the Faculty of Health Sciences of the University of Cape Town (Reference number: 252/2010) (Appendix IV).

#### **3.8.2 Consent**

The participants were identified by the allied health care professionals involved at the Toy Library clinics at the RCWMCH and TBH, who briefly explained the purpose of this study to them. A more detailed explanation of the aims and objectives of this study were then given to the participants by the researcher. No form of persuasion was used to encourage them to participate and they were assured that if they did not wish to participate it would not have any negative effect on the future medical management of their child or family.

Consent was given by the parents for the use of photographs in this dissertation and any other form of publication (Appendix II). All of the photographs, except three, that are presented in this research project are not those of the children of the individuals who participated in the study.

#### **3.8.3 Confidentiality**

The audio-recordings were transcribed as soon as possible after the interview, as confidentiality was a central concern. The audio-recordings were stored on a password-protected computer and transcriptions were kept in a locked filing cabinet in the

Division of Human Genetics and were destroyed once the study was written up and submitted. To ensure anonymity, each participant received a numerical code and this code was used on the interview schedules, audio-recordings, transcripts and spreadsheets.

### **3.8.4 Risk Benefit**

The risk to the participants in this study was the discussion of sensitive information and stressful experiences, which might have evoked suppressed emotions. The researcher was sensitive to the emotional state of the participants throughout the interviews and those participants who felt the need were referred to social or psychiatric services. The researcher ensured that confidentiality and anonymity was maintained at all times. The participants were given the opportunity to discuss and deal with any emotional issues and questions evoked by the initial interview at the follow-up interview.

The long term benefit of this study will be to use the information to improve, if necessary, the health care services and other supportive services offered to parents with children with DS who make use of these services at TBH or RCWMCH.

## **3.9 ASSUMPTIONS**

The researcher assumed that the responses of the participants were honest and a true reflection of their lives.

## **3.10 LIMITATIONS AND STRENGTHS OF THE STUDY**

### **3.10.1 Limitations of the study**

- A major limitation of this study was the small sample size. Due to the time-consuming nature of the interview schedules and the time constraints of this project, only a small number of participants could be included.

- The results of this study will only be able to be generalised to the larger mixed ancestry population attending the services at the RCWMCH and TBH. The results will not be valid for the wider population of parents with children with a preschool child with DS. The reason the results can not be generalised is because a statistically representative sample was not used, purposeful sampling was used. Generalisability is not an objective of this type of research. A small sample size is used as an objective of qualitative phenomenological research in order to have an in-depth understanding of the participants' perspective of DS.
- The data is only representative of one point in time in the participants' lives. This is a limitation, because the parents might experience different needs at different stages of their lives and these changes in the sample were not explored.
- Generally individuals who are willing to participate are those who feel more positive about their child's disability and experience fewer problems in their daily lives. Therefore the data might not be a true reflection of the burden of DS on family life (Siklos and Kerns 2006).
- The researcher has limited interviewing and research experience and therefore may not have had the skills to gather the maximum information during the interviews.
- All the interviews were conducted in a formal setting which could have made the participants feel less comfortable to discuss their emotional issues than if they were in their home environment.
- Even though the researcher is fluent in both languages, it is possible that the data may have lost some of its authenticity when it was translated and slightly edited to make it easier for the reader to understand (Denscombe 2008).
- The paucity of available literature regarding the experiences of parents of children with DS in developing countries, especially SA, made it difficult to find a point of reference and comparison. Therefore information gathered in other countries was used, while exercising caution in generalising between populations from different countries, geographical areas and socio-economic statuses.

### 3.10.1 Strengths of the study

- The researcher was an outsider to the Toy Library clinics and therefore had no vested interest or agenda for personal benefit. The participants also felt more comfortable to freely discuss the negative aspects of their experience with the services than with someone they were accustomed to seeing at the clinics.
- The researcher conducted all the interviews personally. This is a strength because it allowed face-to-face contact with the participants where facial expression and body language could be observed. It also allowed the researcher the opportunity to ask prompt questions which allowed exploration of the aspects in greater depth so that adequate responses were obtained for each question. The researcher is bilingual and fluent in both English and Afrikaans; therefore the researcher was able to conduct the interviews without having to use the services of a translator.
- The open-ended questions asked during the interviews allowed the participants the opportunity to express themselves freely without the limitation of preset categories.
- Audio-recording the interviews allowed a more complete record of the participants' responses than only hand written notes by the researcher.
- The originality of the approach with the use of semi-structured interviews and both quantitative and qualitative methods.
- This is the first study in SA to investigate the experiences of parents with a preschool child with DS.



## Chapter 4

### ANALYSIS, FINDINGS AND DISCUSSION



### 4.1 INTRODUCTION

The data analysis and findings of the research are presented in this chapter. The data are presented in tables and graphs which are followed by a discussion. Where possible, reference is made to the literature to demonstrate similarities and differences in findings of other studies on DS. In total 10 interviews were conducted with mixed ancestry parents of preschool children with DS.

### 4.2 INTERVIEW PROCEDURE

Nine interviews were conducted with the mother and one interview with the father of the child with DS. Five of these interviews were conducted in a private room at the RCWMCH and the other five in a private room at the DSAWC.

All of the interviews took less than an hour to complete. Four of the interviews were conducted in Afrikaans and the other six in English. None of the participants expressed the need to have a follow-up interview. However two participants expressed the need for additional psychosocial support and a referral was made to their respective day hospitals' psychiatric services.

The participants will be referred to by numbers throughout this chapter for ease of reading and to ensure confidentiality. The number refers to the adult who was interviewed. The direct quotes of some of the participants' responses are included to provide the reader with greater insight into their thinking.

### 4.3 SOCIODEMOGRAPHIC INFORMATION OF PARTICIPANTS

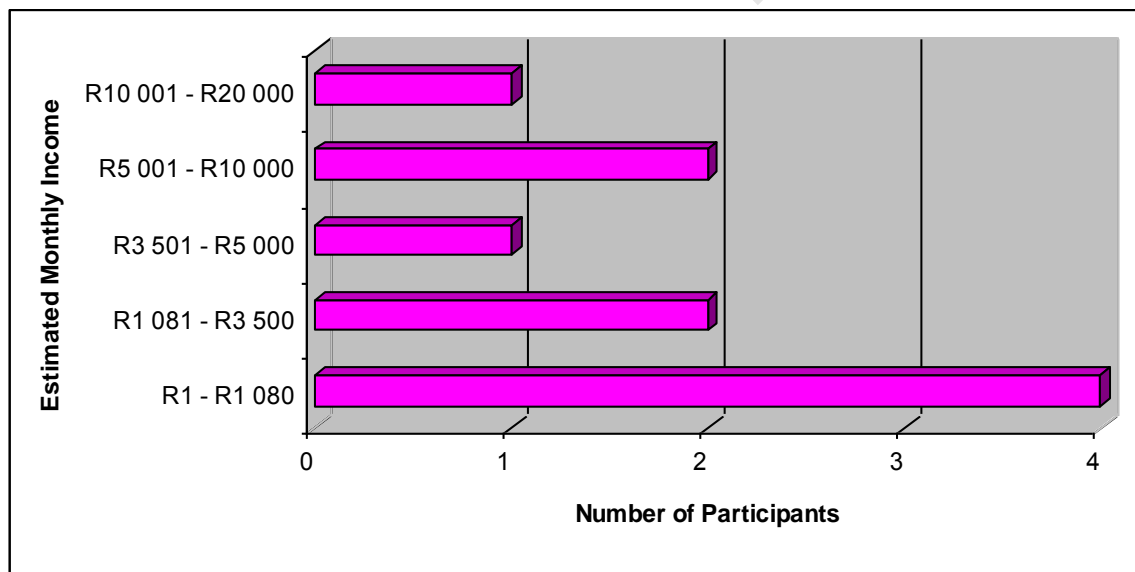
A summary of the sociodemographic information of the participants is presented in Table 4.1.

**Table 4.1: Summary of sociodemographic information of participants**

P. No	Relationship of participant to child with DS	Age of participant (in years) at time of interview	Age of child with DS (in years)	Maternal age (in years) during pregnancy of child with DS	Marital Status	Level of education		Employment status	
						Mother	Father	Mother	Father
1	Mother	34	3	31	Widow	Tertiary	N/A	Full-time	N/A
2	Mother	34	4	30	Married	Gr 11	Gr 12	Self-employed	Self-employed
3	Mother	37	2	35	Informal relationship	Gr 11	Gr 10	Unemployed	Self-employed
4	Mother	46	3	43	Married	Gr 7	Secondary	Unemployed	Part-time
5	Father	33	1	36	Married	Gr 12	Gr 11	Full-time	Part-time
6	Mother	36	6	30	Married	Gr 12	Gr 9	Part-time	Full-time
7	Mother	22	3	19	Married	Gr 12	Gr 10	Unemployed	Unemployed
8	Mother	20	2	18	Single	Gr 10	N/A	Unemployed	N/A
9	Mother	40	6	34	Divorced	Gr 12	Gr 10	Unemployed	N/A
10	Mother	41	2	39	Married	Gr 6	Primary	Unemployed	Full-time

The ten participants included one male and nine females with an average age of 32 years and a range of 19 to 43 years. All the participants were from mixed ancestry and middle to lower socio-economic income levels according to the census data (SSA 2001). The minority of the participants completed high school and only one participant had gone on to complete education at a tertiary level.

The majority of the participants were unemployed and five out of the six participants explained that they were unable to work as they had to care for their child with DS. This finding is similar to an American study conducted by Schieve et al (2010) and is often seen amongst parents with a child with DS. The average household income per month is illustrated in Figure 4.1.



**Figure 4.1: Estimated monthly household income (n=10)**

According to the latest information available from Statistics South Africa (SSA) (2001), the average annual household income in the WC in 2000 was R45 000 (R3 750 per month). The majority of the participants had a monthly income below the average income in the WC. Four of the participants (P3, P7, P8 and P9) relied only on the disability grant that they were receiving for their child with DS. P3 and P9 received

some additional financial support from the father of the child and P7 and P8 were being supported by their parents, the child's grandparents.

A summary of the housing situation for each participant is illustrated in Table 4.2. Figure 4.2 and Figure 4.3 illustrate the houses that some participants live in. All the participants were from lower socio-economic areas or informal settlements. The majority of the participants who made use of the services at the RCWMCH (P1 to P5) had formal housing and the majority who made use of the services at TBH (P6 to P10) had informal housing.



**Figure 4.2: Small government sponsored brick house**



**Figure 4.3: Wendyhouse (wooden dwelling)**

**Table 4.2: Summary of housing situation of participants**

<b>P. No</b>	<b>Area</b>	<b>Housing type</b>	<b>No of Bedrooms</b>	<b>No of Bathrooms</b>	<b>Other rooms</b>	<b>No of occupants</b>	<b>Relationship to participant</b>
1	Mitchell's Plain	Formal house	2	1	Dining room Kitchen Lounge	4	Children
2	Landsdowne	Formal house	3	2	NA	5	Husband and children
3	Mitchell's Plain	Rental house	3	2	Kitchen Lounge	8	Children, friends and friends' children
4	Athlone	House in backyard	2	1	Kitchen	6	Husband and children
5	Newfields	Formal house	3	2	Kitchen Lounge	5	Children and wife
6	Mitchell's Plain	Formal house	4	1	Kitchen Lounge	5	Husband, children and mother-in-law
7	Beacon Valley	Rental house	3	1	Kitchen Lounge	4	Husband and children
8	Strandfontein	Bungalow in backyard	1	0	NA	4	Child, mother and brother
9	Ravensmead	Wendyhouse	1	0	Kitchen	3	Children
10	Wesbank	Government sponsored house	1	1	Kitchen	4	Husband and children

#### **4.4 BREAKING THE NEWS**

##### **4.4.1 Participants' experiences of manner in which news was broken**

The participants were asked a series of questions to establish the circumstances under which they were told about their child's diagnosis of DS. They were asked to describe their experiences of this event. A summary of their responses are indicated in Table 4.3.

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**Table 4.3: Summary of circumstances under which news was broken and participants' initial response**

P. No	Age at Diagnosis	Health care professional who broke the news	Circumstances under which news was broken	Initial emotional response to diagnosis
1	Birth	Paediatrician	Private room with only parents present	"sad"
2	Birth	Midwife	Private room shortly after delivery with only parents present	"scared" "shocking"
3	Birth	Obstetrician	Immediately after delivery in labour ward	"crying" "upset" "sad" "disappointed"
4	Birth	Nursing sister	Private room shortly after delivery with only mother present	"normal" "satisfied" "cry from happiness" (translated)
5	4 or 5 months	Paediatrician	Private room with only parents present	"bit downhearted" "wasn't that difficult"
6	Birth	Medical Geneticist	Private room with only parents present	"glad" (translated)
7	Before birth (5 months gestation)	Genetic Counsellor	Private room with only parents present	"upset" "relieved"
8	Birth	Doctor	Labour ward with other patients present	"was hard" "hurting" "shocked"
9	Birth	Nursing sister	Private room with mother present as well as obstetrician and another nursing sister	"shocked" (translated)
10	Before birth (4 months gestation)	Medical Geneticist	Private room with only mother present	"sad" "painful" "shed tears" (translated)



The majority of the participants described an initial feeling of shock or sadness when they were told that their child had DS. These findings are consistent with the findings reported by previous researchers (Noble 1993; Burke and McDaniel 2001; Skotko 2005).

Similar to what has been reported in the literature, the majority of the participants only found out their child had DS postnatally (Skotko 2005). These eight out of the ten participants (P1, P2, P3, P4, P5, P6, P8 and P9) were asked whether they suspected anything different about their child before they were told of the diagnosis. Only P1 and P6 suspected that there was a problem. P2 and P3 were told shortly after birth and only saw their child for the first time after the news was broken. P4, P5, P8 and P9 had no idea their child had DS prior to when the diagnosis was made. P8 felt her daughter with DS had the same facial features as she had. P9's experience with the birth of her first child contributed to her disbelief that her second child had DS. When asked whether she could see anything different from her child at birth she responded:

*"I couldn't actually, because it was almost the same with my oldest one, but I was longer in labour with the oldest one and her forehead was also flat and she had a flat face, but with time it came right. So I thought maybe it can be the same with her (child with DS) and that after birth she will come right."* (Translated)

Two of the participants (P7 and P10) received a prenatal diagnosis of DS. P7 was offered testing, because of soft markers that were seen on the ultrasound scan at five months gestation. P10 was offered diagnostic testing, because she was of AMA and because they detected foetal anomalies on the ultrasound scan at four months gestation. Neither of them wanted to terminate the pregnancy for religious reasons as P7 was of Muslim and P10 of Christian belief. P7 was initially counselled and told that they suspected her child to have a severe chromosomal anomaly and that she would not live very long. After prenatal diagnostic testing, P7 was relieved when she was told her child had DS and not this more severe problem. P7 described this event as follows:

*"They first told me they found all the problems on the scan and then they told me that first it was T something, that the child won't live long...and then I went for the amnio*

*test and they came back to me and said it's T21. I was first devastated when they told me the child is not going to live very long, like a day or a month or a year. I was actually relieved when they told me it is DS and not that type of baby."*

Like P10, P4 was also of AMA while expecting her child with DS. She was however not offered any prenatal screening or testing as she only initiated antenatal care at a late stage, after 20 weeks gestation. This is a common occurrence in SA as pregnant women of lower socio-economic status lack the knowledge of the benefits of booking a pregnancy early as was the case in this instance. P4 also had financial difficulties which have also been reported to play a role (Abrahams et al 2001; Myer and Harrison 2003).

Even though the majority of the parents found the news itself traumatic, they were satisfied with the manner in which they were told that their child had DS. Eight out of the ten (P1, P2, P4, P5, P6, P7, P9 and P10) would not have wanted to be told in any other way. These findings differ from the findings of Skotko and Canal (2004), Hedov (2002) and Muggli et al (2009) who respectively reported dissatisfaction amongst American, Swedish and Australian parents.

Five of these participants (P1, P5, P6, P9 and P10) experienced this event more positively than the others, because they felt supported by the health care professional who gave them the news. These health care professionals took time to sit down with the parents and explained what DS is and what future management would entail. They also scheduled follow-up appointments with the parents to discuss any further questions regarding their child with DS. Muggli et al (2009) reported similar findings in a study conducted in Australia.

Skotko (2005) suggested that religion played an important role in how American parents coped with the news. The current research supported this finding. P2, P3, P4, P5 and P10 were found to deal better with the news due to their religious beliefs. P2, P4 and P10 were of Muslim belief and P3 and P10 were of Christian belief. Both P4 and P5 stated that they were "*happy*" when they found out their child had DS and did not

struggle to accept the news due to their belief that these children will carry them into heaven. P5 explained their beliefs as follows:

*“A DS child, or any child that is not of full mental ability or physically disabled at birth, are promised to go to Jannah. Jannah is what we call heaven. Our religion says that they are going to be waiting for us no matter how long. So it makes things a lot easier for us.”*

P2, who is of Muslim belief, and P3, who is of Christian faith, both believed that their child was a gift from God and therefore found it easier to accept their child with DS.

The current study found that having been in close contact with someone who has DS contributed to experiencing this event more positively. P4 and P5 both had siblings with DS and P6 had a friend with a daughter with DS. These participants made the following comments:

P4: *“I had a sister like that. We were all happy for her (child with DS).”* (Translated)

P5: *“For me it wasn’t that difficult, because I have a DS brother.”*

P10: *“My friend had a daughter with DS, so it was actually easier for me.”* (Translated)

Two out of the ten participants (P3 and P8) expressed intense dissatisfaction with the manner in which they were told that their child had DS. Both participants were told in the labour ward with other patients present. These findings are similar to those reported by Garwick et al (1995) and Skotko (2005) amongst mothers in the USA.

P3 was told while she was still in pain from the Caesarean section and P8 described being *“out of it”* and *“tired”* when she was told. P3 experienced this event the worst and gave the following description:

*“And the doctor just said to me your baby is not normal, but that wasn’t the words that I was expecting to come out of a professional’s mouth. Because I don’t know, not normal can mean anything. I just burst out crying and then I was upset, because nobody came to me and said to me what was wrong. Your mind can wander off with you. What is not normal? I mean he could have been disfigured.”*

Both participants stated that they will never forget the way they were told and how they felt at that moment. This phenomenon has also been reported amongst Australian mothers in a study conducted by Muggli et al (2009).

These two participants were asked how they would have preferred to be told that their child had DS and responded:

P3: *"They could've at least waited until they closed me up and had pushed me into a room where I semi-recovered and gained my consciousness and said: 'Listen, I don't know if you've heard of the term DS', or just show me a picture, then I would have been fine with it."*

P8: *"I would prefer her taking me to a place where it was more private. Where she could tell me."*

### **4.4.2 Experience of conveying news to family and friends**

Each participant was asked whether they had conveyed the news of their child's diagnosis to anyone when they had found out. They were also asked to describe the reaction of these individuals and how they experienced having to tell someone that their child had DS. Their responses are indicated in Table 4.4.

**Table 4.4: Participants' experience when conveying the news about DS to family and friends**

P. No	Individuals news of diagnosis was shared with	Reactions of family and friends	Reactions of partner	Participants' experience of having to convey news of child's diagnosis
1	Husband Mother Family	"They all thought that it's a generation thing and my sister-in-law still thinks it's a genetic thing...like from a generation to a generation... They think that it's something to do with us."	"He took it a bit harsh. He said it couldn't have happened."	"...beforehand I tell people so that the people don't wonder why the child is delayed. So that would make me feel better."
2	Sister Boyfriend Family	"She (sister) said: 'No man, there is nothing wrong with her.' "...seeing me, the mother, handling the situation so well made them happier."	"I don't really think that he ever doubted her (child with DS) or anything, but for him it was very hard."	"I was mostly scared, but more happy, because in our religion it is that they are special. So we could tell friends and relatives that it was a blessing."
3	Mother Grandmother Best friend	"She (best friend) said to me she's shocked." "They (family) all accepted him, because in my granny's far back family, they had a little DS." "My daughter, she was a bit shocked."	"My boyfriend was just upset...it was like the whole mountain was falling on top of his shoulders, but because I was in pain as well, he kept being strong, but when he went home I knew the reaction. He was going to kick things around and question himself. "	"The second day I didn't feel anything, because I was strong enough to accept it. I was proud of myself to say that and the second day I could have told people that I've got a DS."

4	Sister-in-law Husband	"They were content. They were a little uncertain, because they have never had something like that in their family. Everyone was happy and fond of (child with DS)." (translated)	"Uncertain." (translated)	"I wanted to cry not because of sadness, but because of happiness." (translated)
5	Family	"Some of my family, like my cousins, didn't know how to react to me explaining it to them so easily. Because of my brother everybody accepted and loves him (child with DS)."	"My wife wasn't so happy, but after a few days she came to the party."	"It wasn't a problem. Really it wasn't a problem."
6	Family	"The family was very supportive." (translated)	"supportive" (translated)	"It's not easy to tell someone I have a child with DS, because people don't understand. People don't know how to handle a child with DS... You get people that you are comfortable with and then you get people who you think are ignorant and decide not to tell them. Up until today I get hesitation to say I have a child with DS." (translated)
7	Mother Family	"She (mother) was surprised, not surprised, she was actually quite happy, because she likes Downs children. So it wasn't like a problem." "We got through it and it brought us closer together."	"Had a lot of support."	"I was okay with everything. At first I was a bit upset, but I overcame it."

8	Paternal aunt Father	"My aunty wasn't happy, not happy, but she felt bad for me. My father also. He actually blamed the doctors and he blames my mother because of my mother not supporting me throughout my pregnancy."	NA	"I can't even explain right now. It was very difficult. Like for me it was like I had to hide it away. I didn't want people to know about it, because people are going to make fun and I wasn't up for that."
9	Husband Family	"It was a shock for them (family) as well to tell you the truth. They all love her (child with DS) and care about her." (translated)	"He couldn't accept it." (translated)	_____
10	Best friend Best friend's husband Family	"I told them (friends) and they stood by me until I gave birth and up until today still." "It was a very difficult time for the family in the beginning." (translated)	"sad" (translated)	"I was delighted to tell other people. I have a smile on my face when I tell them. Every now and then the tears start rolling as I tell them and sometimes there is one or two who cry with me." (translated)

All of the participants shared the news of their child's diagnosis shortly after they found out themselves. The majority did not find it difficult to share and experienced it positively. P1 and P3 stated that it made them feel better to speak about it. P2 found it easy, because of her religious beliefs. P4 and P10 were happy to share the news, but both described "*shedding tears*" when doing so. P5 and P7 did not find it difficult to speak about their child's diagnosis. Two out of the ten (P6 and P8) found it difficult to share the news, not because they found it difficult to come to terms with the diagnosis, but because of the fear of how the people would react towards their child with DS.

The majority shared the news of the diagnosis with their family members who reacted in different ways ranging from feelings of blame, denial and disbelief to shock and sadness. Two of the participants (P4 and P5) highlighted that some of the family members did not know how to react towards them.

Even though the majority of the participants' family also experienced this event as a shock, they were very accepting towards the child with DS and were an invaluable resource of support to the parents. This finding supports the findings reported in the literature by Buckley (2002) as well as Siklos and Kerns (2006).

An interesting phenomenon was the perception of the participants that they coped better with the news than their partners. The majority (P1, P2, P4, P5 and P9) of the participants described their partners as experiencing feelings of denial, anger, guilt, unhappiness and sadness. P9's husband did not want to accept the diagnosis at all. These emotional reactions have previously been described amongst a group of SA parents with a child with DS by Steenkamp and Steenkamp (1992).



## 4.5 HEALTH CARE AND SUPPORTIVE SERVICES

### 4.5.1 Participants' satisfaction with counselling services

All of the participants were asked whether they recalled receiving counselling regarding DS and the causes thereof. Three out of the ten participants (P1, P2 and P8) did not recall receiving any counselling. P2 felt they had received enough information from the genetic doctors and therefore further counselling was unnecessary. P8 was offered counselling, but did not want counselling shortly after the birth of her child with DS. When asked if P8 would have liked counselling at a later stage she responded:

*"I think I do, because just to explain to people how I felt about having a child with DS. Having all those feelings bottled up inside and keeping it. It would've been best to speak about it."*

P1 did not acknowledge ever receiving any form of counselling or specifically genetic counselling. She said she was advised to contact someone, but never did due to time constraints. She did however mention that she had an "interview" with a genetic counsellor when her son with DS was three years old. She described the interview as follows:

*"She told me that it came from the mom's side, because the dad doesn't give the sperm or the whatever. It was a genetic fault and that was actually bad, because coming from you, from the female side that I took a bit hard."*

The experiences of the other seven participants that did recall receiving counselling are summarised in Table 4.5. The majority were satisfied with the counselling that they had received. These findings are consistent with the findings reported by Michie et al (1997) amongst couples attending the genetic counselling services in London, as well as the findings reported by Stadler and Mulvihill (1998) amongst American Breast cancer patients.

**Table 4.5: A summary of how the participants experienced the counselling services.**

P. No	Age of child when counselled	Counsellor	Hospital	Description of counselling received	Positive aspects?	Negative aspects?
3	Few days	Nursing sister	MMH	"Just the basics of what to expect in the future, but nothing about the causes."	None	"She was talking more about her brother, because she's got a brother that's DS. Explain to me what am I going to expect, not what your brother has been through and where he is at the moment."
4	Few days	Doctor	HPP	"She explained everything to me. What it is, how I must be and what I must keep an eye on. What I must actually take note of regarding (child with DS). It was very helpful." (translated)	"The best was that she told me what I must keep an eye on regarding DS and what I must do if this or that happens." (translated)	None
5	Five months	Genetic Doctor	RCWMCH	"It was very well done."	"Everything so far they told us, it works."	None
6	Few days	Genetic Doctor	GSH	"We received complete counselling for those six weeks that he (child with DS) was in hospital. We actually saw the doctor, two or three times, and she came to speak to us to make us comfortable with DS." (translated)	"Knowledgeable. I learned a lot." (translated)	"They actually told me for those six weeks that he was not going to make it until the next day. I didn't like that at all." (translated)

7	Before birth	Genetic Counsellor	GSH	"Very helpful."	"She made me understand."	None
9	One month	Genetic Doctor	TBH	"I accepted it as the doctor told me and explained it and I just lived with it." (translated)	"The doctor told me it was not my fault and also not his (the father's) fault; because I felt it might have been my fault that this happened. But when she explained that I found peace." (translated)	None
10	Before birth	Genetic Doctor	TBH	"For me it was something good that I have never experienced before. She explained to me every time and that made me feel good and everything." (translated)	"For me it was the information." (translated)	None

Five out of the seven (P4, P5, P6, P7 and P10) found that the most positive aspect of the counselling session was the information they had received and the knowledge that they had gained. One participant (P9) valued the counselling she received as it reduced her feelings of guilt and self-blame. These findings are similar to that reported by Collins et al (2001) amongst a group of Australian parents of a child with CF or DS.

Out of the seven only P3 experienced the counselling she received negatively. The participant felt that she wasn't given enough information on DS and what to expect in future. This could be attributed to the fact that P3 was the only participant counselled by a nursing sister and not a doctor or a health care professional trained in genetics.

Three out of the seven participants (P3, P6 and P9) would have liked something different about the counselling they received. When P3 was asked how she would have liked the counselling to be different she responded:

*"I wanted her, like I said, to go into more detail, because I didn't understand DS...How the chromosomes work, what happened during the process of my pregnancy, but nothing like that was explained to me."*

P6 felt she would have benefited more from the counselling if the counsellor had waited until her child with DS was healthy. She felt that at that stage she was more worried about her child getting better than to hear about DS. P9 was satisfied with the counselling she received, but would have liked a follow-up appointment with the doctor. She felt one session was not enough. Collins et al (2001) reported similar findings amongst Australian parents of a child with CF or DS.

### **4.5.2 Participants' experience of health care services at RCWMCH and TBH**

The participants were asked questions regarding their experiences with the health care services at the RCWMCH or TBH. Table 4.6 gives a summary of the services each participant made use of as well as their experience thereof.

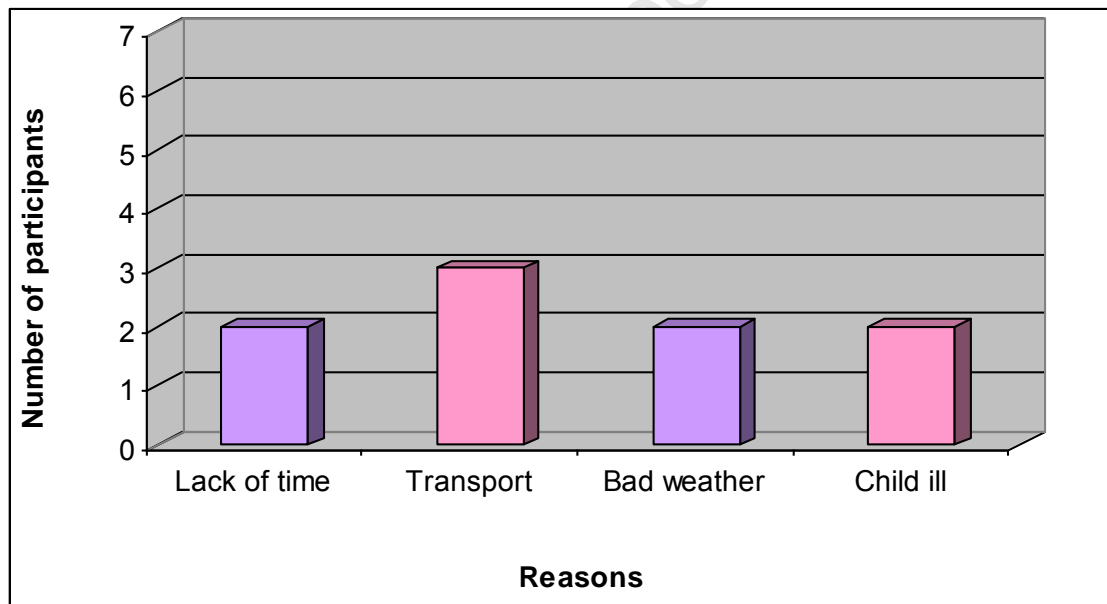
**Table 4.6: Summary of participants' experiences of health care services**

P. No	Attending hospital	Attending clinics	Transport	Cost of transport	Experience of services?
1	RCWMCH	Toy Library Cardiac Paediatric	Own motor vehicle	R50	"It's been well."
2	RCWMCH	Toy Library Developmental ENT	Own motor vehicle	R25	"Very good. No complaints."
3	RCWMCH	Toy Library Speech Cardiac Eye Occupational Therapy	Own motor vehicle	R50	"Excellent. I've got no problems."
4	RCWMCH	Toy Library Developmental Dietician	Taxi	R30	"Very good." (translated)
5	RCWMCH	Toy Library Developmental Speech Genetic	Own motor vehicle	R40	"Everything has been brilliant."
6	RCWMCH and TBH	Toy Library Respiratory	Own motor vehicle	R30	"It's a public hospital so it was okay." (translated)

7	RCWMCH and TBH	Toy Library Genetic Cardiac ENT Endocrine	Taxi	R20	"Everything is okay, it's just I had a problem with them when the folder went missing. They have improved, but sometimes there have poor service, like the pharmacy, you sit quite a long time there."
8	TBH	Toy Library Speech Genetic ENT Physiotherapy	Bus	Unknown	"They are doing everything fine."
9	TBH	Toy Library Cardiac Eye	Walks	No cost	"It's okay, I am content with their services." (translated)
10	TBH	Toy Library Eye Genetic	Taxi	R20	"For me it is yes the clinics help, but sometimes there are problems with the sisters." (translated)

P1, P2, P3, P4 and P5 were all attending the health care services, including the Toy Library at the RCWMCH and P8, P9 and P10 the services at TBH. P6 and P7 were making use of the health care services at the RCWMCH, but preferred attending the Toy Library at TBH. Half of the participants owned their own motor vehicle to use as transport to the hospital, while four participants made use of public transport. P9 could not afford the costs involved with making use of public transport and therefore had to walk to the hospital to attend her child's appointments.

When the participants were asked if they found it difficult to attend all the appointments at the hospital, seven out of the ten (P1, P2, P4, P5, P6, P7 and P9) sometimes struggled to make it to their appointments. Figure 4.4 indicates their reasons for missing a few of their clinic appointments.



**Figure 4.4: Reasons for missing clinic appointments (n=7)**

Children with DS often have to attend the hospitals for appointments at a variety of clinics due to their unique health concerns that are associated with their condition. For this reason the parents find themselves having to attend the hospital almost every month and as indicated in Figure 4.4, sometimes struggle to find the time, especially those

parents who are working. Their lack of transport to attend these clinics was directly related to their financial status. Some parents could not afford the costs of public transport.

Children with DS have lowered immune levels and get ill more easily than other children. They are often hospitalised when they are ill and during this time they miss appointments at the specialised clinics. P4 and P9 did not go to their appointments in bad weather, because of their fear that their child would get ill. P9's only means of transport was walking to the hospital, which she was unable to do in rainy weather.

All the participants felt the services were suitable for their children with DS and they all felt that their health concerns were being adequately addressed. The participants only making use of the services at the RCWMCH (P1 to P5) were much more satisfied with the health care services than those who made use of the services at TBH as well. Eaves et al (1996) and Yam et al (2005) also reported that the majority of a group of caregivers in Canada and Hong Kong respectively felt satisfied with the services. However Wexler et al (2009) found that caregivers of individuals with DS in Israel were not satisfied with the health care services and felt that their needs were being inadequately addressed.

P6 had complaints about the administration of the hospital and the long queues at the pharmacy of the RCWMCH. P9 had similar complaints regarding the long queues. Both P9 and P10 complained about the attitude of the nursing sisters at TBH. They felt the nursing sisters did not have any understanding of what children with DS are like and described the sisters as being unhelpful and inconsiderate.

When the participants were asked if they had any recommendations on how to improve the services, the majority were content with the way things were and didn't have any suggestions. P5 felt that he would have liked it if his child with DS could have received individual speech therapy as well, because he was very concerned about his child's delayed speech. P7 felt the pharmacy at the RCWMCH should improve their services



and that the hospital should do something to make the clinics less crowded. P9 felt that the hospital should give preference to the parents with a child with DS.

P9: *“All I want is that they should give them preference so that I don't have to wait when I get to the hospital, because it's just a short time that they will sit still.”*

P10 felt that the nursing sister should be more helpful and considerate.

P10: *“They (nursing sisters) can at least phone the doctor and tell the doctor your patients are here...They just sit there and talk and don't tell the doctors.”* (Translated)

## **4.6 SOCIAL SUPPORT**

### **4.6.1 Participants' experience of social support**

The participants were asked who they felt supported them the most and how they experienced the support they were receiving from their family and partners, if applicable. Table 4.7 provides a summary of the participants' responses.

**Table 4.7: Participants' experiences of social support from family and partners**

P. No	Who supports you the most?	Experience of support from...		Are you receiving enough support?
		Family members	partner	
1	"Myself"	"My mom helps me sometimes if I have a problem. If it's school holidays that the children don't have anywhere to go then I'm forced to drop them at my mom, but it's actually not that they want to look after the kids, but I'm forced to do that."	NA	"Not actually"
2	"My husband and my family"	"I would say that getting advice from your older siblings, it's amazing"	Supportive	"Over enough"
3	"My boyfriend"	"My mother supports a lot as well."	"His father is standing by me up until today still."	"Yes"
4	"My family" (translated)	"Very good. No one has ever said anything nasty about her. They all love her." (translated)	Supportive	"Yes, you must be content with what you get." (translated)
5	"My wife"	"The family is brilliant."	"We support each other with everything."	"Yes."
6	"My husband" (translated)	"It is actually just my mother-in-law that helps" (translated)	"He's there when I'm not there." (translated)	"I think it is enough." (translated)

7	"My husband"	"I always let my mommy know first. She helps financially."	"I get lots of support from him."	"Yes"
8	"My mom"	"Yes, but it's just that I feel that because I am her mother I should do more, which I cannot at the moment because I am not working."	NA	"Yes"
9	"My sister-in-law"	"She has been with me since day one. If I have to go somewhere then she tells me to bring the children to her." (translated)	"He can put in a bit more." (translated)	"Yes"
10	"My husband, my parents and his (child with DS) godparents"	"For me, I can feel the love is there, they are not pushing me away because of (child with DS's) condition." (translated)	"He supports me." (translated)	"Yes"

## CHAPTER 4: ANALYSIS, FINDINGS AND DISCUSSION

The majority of the participants (P2, P3, P5, P6, P7 and P10) felt they were receiving the most support regarding their child with DS from their partners. All of the participants, except P1, felt that they were receiving an adequate amount of social support. P1 gave the following response when asked if there is anyone she felt always helped her:

*“No, I need to be everything. I need to be the dad, the mom, the financial aid, everything.”*

When asked what supports she felt would help she suggested the following:

*“Maybe people you could go to and have their kids play with your kids. Something like that just for a break, because I don’t have that.”*

Even though P1 was regularly attending the Toy Library at the RCWMCH she had no information regarding other available support groups and family get-togethers they often organised.

P8 felt her friends were not interested in her child and did not support her with her child with DS. She felt that their support would be helpful. Previous studies have also indicated that parents feel it is important for them to be supported by their friends (Ross and Deverell 2010).

Two out of the ten participants (P8 and P10) identified two other important sources of social support. P8 felt that the support group she was attending was helping her cope with raising her child with DS. P10 felt that belonging to a church and the congregation accepting her son with DS, made coping a lot easier. These findings are similar to the findings reported by Buckley (2002). Both of these resources provide the parents with a sense of belonging and not being excluded from the community due to their child with DS.

Two of the participants (P5 and P10) highlighted an important aspect regarding the effect of interacting with other parents and their children with DS had on the coping of the participants. These participants perceived their children as doing better than the others and this seemed to create a more positive attitude towards their child with DS. P5 described his child as follows:

*“Compared to others he is very good, he is a year and a half and he has started to talk which the other boy who lives close to us cannot do. He can’t sit properly and he can’t say anything.”*

P10 commented on feeling grateful that her child was not like the other children with DS.

*“Why is it that that child is like that and mine isn’t? I always say thank God that my child does not have that many problems.”* (Translated)

#### **4.7 EXPERIENCES OF PARENTS WITH A PRESCHOOL CHILD WITH DS**

##### **4.7.1 Discussion of impact of child with DS on participants’ everyday life**

The participants were asked to comment on how having a child with DS has affected their daily lives and their relationships with their partners and other children.

The majority of the participants felt that having a child with DS had not caused any major problems in their daily lives. Four out of the ten (P1, P5, P6 and P10) identified a few major difficulties. P1 felt that it was difficult for her to find the time to spend with her child with DS to teach him things properly. P5 said that they found it difficult to get their child to sleep at night. When P6 was asked what she found as a major difficulty she responded:

*“To leave him with my mother-in-law when I go work. To leave him with her and I know she is not actually well every day then I wonder if she will be able to take care of him.”* (Translated)

P9 was experiencing major difficulties due to the circumstances she was living in. Her landlady did not approve of her child with DS and had told the participant to keep the child inside the house.

*“(Child with DS) was told by the landlady that she is not allowed to play outside and make a noise or sing loudly, because then they have a problem. Now I must keep her in the house and the wendyhouse is small.”* (Translated)

P9 admitted experiencing a lot of emotional problems, because of this situation and admitted feeling depressed at times.

Half of the participants (P2, P3, P5, P6 and P7) did not feel they experienced any emotional problems. The other five participants mostly reported feeling “*sad*” and “*lonely*” at times.

P1 and P2 felt lonely at times and missed having the support from their child’s father.

P1: “*You don’t have anybody to share the pain and things with.*”

P8: “*The sad thing is you get those fathers with the mothers that are so fond of their child and caring towards their children. Now I don’t get that, I am the one struggling.*”

These findings support the findings of Erickson and Upsher (1989) who indicated that American mothers’ perception of the caretaker burden was lighter when the father participated in taking care of the child and provided them with emotional support..

P4 often experienced feeling “*sad*”, not because of having a child with DS, but because of her financial situation. She wanted to give her children all that they wanted, but felt she could not because there was not enough money for her to do so. P8 expressed similar feelings regarding her finances and it inhibiting her to do more for her child. Cuskelly et al (2008) emphasised the importance of adequate financial resources in the coping process of parents in the USA which are once again shown in the findings of this study.

### **4.7.2 Discussion of impact of child with DS on marital relationship**

The eight participants (P2, P3, P4, P5, P6, P7, P9 and P10) who were married, in a relationship or had been married were asked how having a child with DS had affected their relationship.

For most of the participants it had either no effect or they felt it made their relationship stronger. This is consistent with the findings reported in the literature by Van Riper (2002) amongst parents in the USA. P2, P4 and P6 felt having a child with DS had absolutely no effect on their marital relationship. P3, P5, P7 and P10 felt having a child

with DS had a positive effect on their relationship and described them as being “*happier*” and “*closer*”.

P9 was the only participant who felt that having a child with DS had affected her marital relationship negatively. She and her husband got divorced due to the increasing problems in their relationship after their daughter with DS was born. When P9 was asked to explain what the reason for the divorce was she responded:

*“He actually said that I did not give attention to him, because I was giving attention to (child with DS), but (child with DS) was always sick. Really, I was more in hospital than I was at home. And it happened when I was in the hospital that time that he had an affair.”* (Translated)

The finding of the participant separating from her husband, due to him feeling neglected, is similar to that reported by Sari et al (2006) amongst Turkish couples with a preschool child with DS.

#### **4.7.3 Discussion of other children’s attitudes towards their sibling with DS**

Nine of the participants were asked to comment on their other children’s attitudes towards their sibling with DS and to comment on their relationship. P8 were not asked these questions as she only has a child with DS.

Six (P2, P3, P4, P7, P9 and P10) out of the nine had no problems with their children’s attitudes toward their sibling with DS and felt they got along well with each other. P2 described their children’s reactions towards their sibling as “*normal*” and P2, P4 and P10 commented on how much their children loved their sibling with DS. P9 felt that her other child helped a lot with caring for her sibling with DS. . These findings support the findings reported by Cuskelly and Gunn (2003) who compared the adjustment of a group of siblings of a child with DS with a comparison group of siblings of children who were developing typically.

Three of the participants (P1, P5 and P6) experienced some troubles with their other children's attitudes towards their sibling with DS. Even though P1 described the interaction between her children as "*normal*", she mentioned feelings of jealousy and the perception of favouritism by their mother at times. When asked about the children's feelings she responded:

*"They don't think he's (child with DS) different, but sometimes, because I treat him, not special but I give him more attention they feel that (child with DS) gets the upper hand."*

P5 had another child with autism who seemed to imitate everything his sibling did who has DS. The participant described the relationship as follows:

*"The brother, the autistic one, is very jealous, because (child with DS) is getting a lot of attention. So it's getting him to also do things, because it's like he is also looking for the compliments, because of (child with DS)."*

This behaviour of imitation and attention-seeking has also been reported by Sari et al (2006) in a group of Turkish siblings of children with intellectual disabilities.

P6 felt she was going to have a problem with her teenager, because he did not have enough patience with his younger sibling with DS. When asked how he reacted towards his sibling, P6 responded:

*"He accepts his brother has DS, but he doesn't understand that Downs can't laugh, talk and understand, because he can't talk full sentences yet. I think I am going to have problems with him, because he is a teenager and he doesn't have that patience with the baby."* (Translated)

#### **4.7.4 Participants' self-reported experience of child with DS**

Each participant was asked to describe the personality and how they experienced the behaviour of their child with DS. They were also asked to comment on what have been the most unpleasant experiences for them regarding their child as well as the positive experiences. A summary of their responses are presented in Table 4.8.



**Table 4.8: Participants' experiences with having a child with DS**

P. No	Description of personality	Description of behaviour	Negative experience	Positive experience
1	"Normal."	"Naughty." "...don't listen."	"Taking him to hospital and when he was sick that time I didn't have any support from my family"	None
2	"She's always got a smile."	"She has a temper, but not terrible behaviour..."	"Just coming to the hospital everyday and thinking what's going to happen now? What are the doctors going to tell us now?"	"It makes you love the next child with DS whether the child is black or white. Just to have that love for children, not for yours only, but for others as well."
3	"Very friendly." "...likes to be free." "...likes people around him."	"Very well." "Hyper..."	"Nothing, nothing at all. That is now the honest truth."	"(Child with DS) brings so much joy in my life and people remember him wherever we go."
4	"...quiet around strangers..." "Very lovely." "Shy." (translated)	"Very good, but naughty like any child." (translated)	"I am just a little unhappy if she (child with DS) is sick the day, because then there is no playing or laughing in the house, because everyone feels sorry for her. I am very stressed when I see she is getting sick. I am just afraid that I am going to lose her." (translated)	"She is your happiness. The people love (child with DS) and I can't see that they have anything against her." (translated)
5	"Flustered." "Too friendly." "...interacts well with anybody..."	"Very friendly."	"Sitting in hospital and to sleep on the floor in hospital. Not a good experience, especially last year...he had a drip and they put plasters on his hands. He had tubes and stuff in his nose and he had an oxygen mask on. It wasn't nice."	"We've got a baby for longer. We've got a baby for ever."

6	"...fun..." "...not a child that hangs out with other people." (translated)	"Behaves in public, but not at home." "Naughty." (translated)	"When he was sick, I think that was the most unpleasant. So if he starts getting a cold we start to panic, because we are scared that it's going to get critical again." (translated)	"DS actually taught me to respect other disabilities and to take note. I think that is positive, because it actually makes me human to not be nasty to other people." (translated)
7	"...friendly..." "...sometimes have her moods."	"Very calm."	"It's just when she is in hospital I feel very, not sad that I have to sit in the hospital, but I'm very, not depressed, but I don't like it when she is sick. I am always worried she is going to get sick."	"They bring lots of joy to your life. She brings a smile to you any moment anytime of the day."
8	"Very stubborn." "Very friendly." "...comfortable with anyone."	"Normal."	"There is nothing I can think of."	"Just to take care of her as a DS, because I believe that God had a purpose for me."
9	"Very cute when she was little...now that she is a little older she is getting stubborn" (translated)	"Aggressive." (Translated) "Hyperactive." (translated)	"It was not unpleasant yet"	"I am happy I have (child with DS), because (child with DS) has come to make me stronger. If I did not love (child with DS) and I went through the divorce I would not have made it. She was my pillar of strength." (translated)
10	"Lovely child." (translated)	"Very good." (translated)	None	"We are more happy since we've had (child with DS) and there is always laughter in the house." (translated)

## CHAPTER 4: ANALYSIS, FINDINGS AND DISCUSSION

The majority of the participants experienced the personality and behaviour of their child with DS positively. They most often used the word “*friendly*” to describe their child’s personality. The parents often described their child as being “*naughty*”, but felt this was normal naughtiness like in any other young child’s behaviour. P1 and P10 were the only participants who felt their children exhibited bad behaviour. These findings are similar to those reported in the literature by several researchers (Wishart and Johnson 1990, Myers and Pueschel 1991, Cuskelly and Dadds 1992).

Four out of the ten participants (P3, P8, P9 and P10) felt there was nothing unpleasant about having a child with DS. The majority of the other participants (P1, P2, P4, P5, P6 and P7) felt that the only unpleasant thing about having a child with DS was when the child was sick and had to be admitted to the hospital. The experience of seeing their child in hospital and the fear of losing their child made this the most unpleasant experience for them.

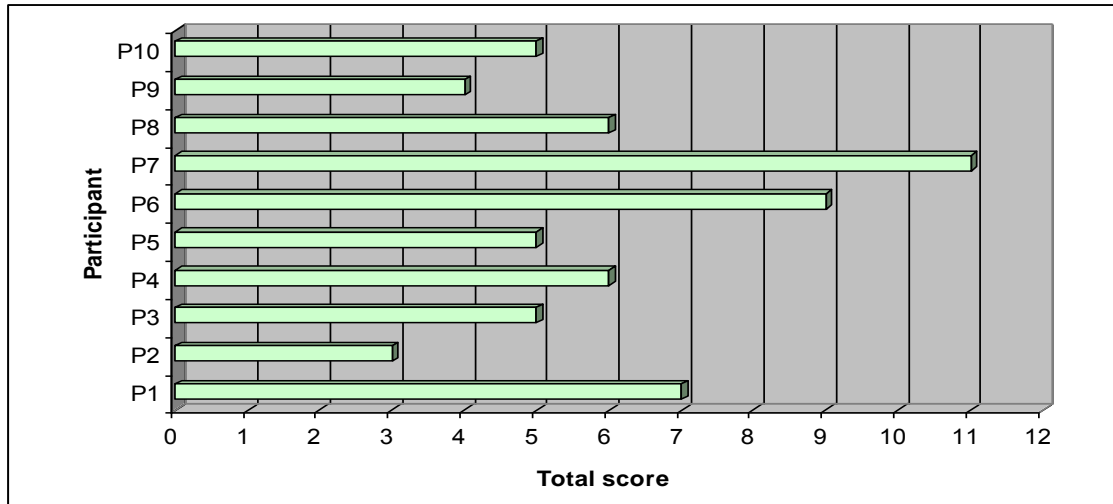
All of the participants, except P1, felt that there were definite positive aspects regarding their experience of having a child with DS. P2 and P6 commented on their children teaching them to love and respect other people even if they had disabilities. Four of the participants (P3, P4, P7 and P10) felt that having a child with DS had brought joy and happiness to their lives and the lives of their family. P5 felt it was a positive thing, because they would always need to be looked after. P8 felt that having a child with DS had been a positive thing in her life, because it gave her purpose. P9 felt that her child had made her strong enough to handle other crises and helped her through the divorce.

Overall the participants felt that the positive aspects of having a child with DS outweighed the unpleasant experiences that they had had. These findings are consistent with those reported amongst American parents by both Abbott and Meredith (1986) and Van Riper (2007).

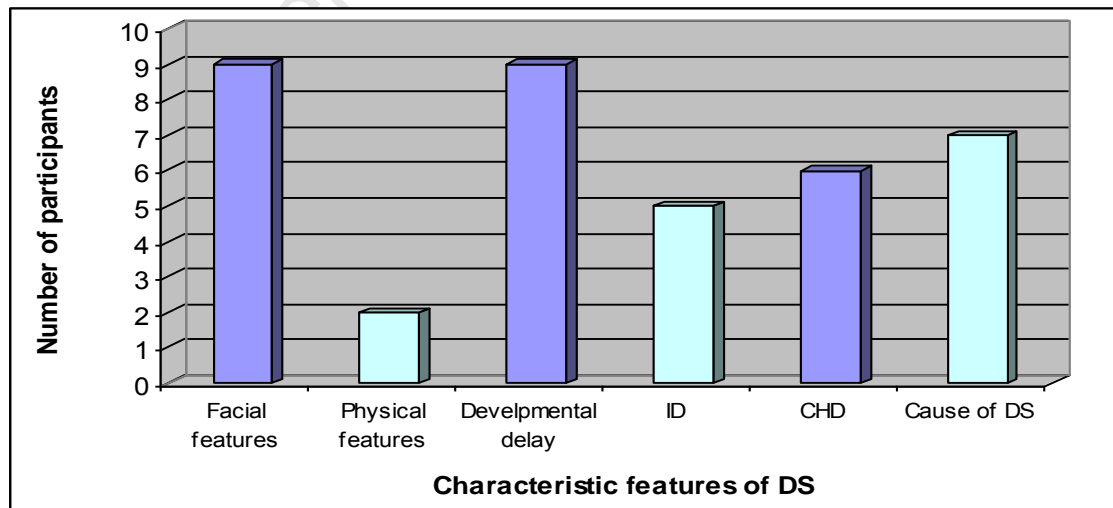
#### 4.8 LEVEL OF PARTICIPANTS' GENETIC KNOWLEDGE OF DS

##### 4.8.1 Discussion of participants' level of understanding of cause and features of DS

The participants were asked to explain what they understood regarding the characteristics of DS and what caused the condition. Figure 4.5 and Figure 4.6 illustrates their level of understanding.



**Figure 4.5: Measurement of knowledge of characteristics and cause of DS for each participant**



**Figure 4.6: Level of understanding of causes and features of DS**

## CHAPTER 4: ANALYSIS, FINDINGS AND DISCUSSION

All of the participants, except P2, could recall the facial features of DS. When describing the features, three out of the ten participants (P2, P3 and P4) lacked the ability to recognise these features in their own children. They made the following comments.

P2: *“But she doesn’t completely look like a child with DS. She’s growing now and it’s almost like you don’t see that features like you know.”*

P3: *“You know some days he looks normal and then the next day it is just the eyes.”*

P4: *“Everyone did not want to believe she is DS, because she doesn’t look DS.”*

(Translated)

When asked to describe the features of DS, only two out of the ten participants commented on the physical features, other than the facial. These other physical features include hypotonia, sandal gap, single palmer crease and shorter length of individuals with DS.

Only half of the participants acknowledged that children with DS have an intellectual disability. When asked if they believed their child would be able to cope in a mainstream school; P2, P3, P7 and P8 felt that their child would do fine in a mainstream school. The attitudes regarding schooling for children with disabilities are rapidly changing and parents are being encouraged to place their children in mainstream schools (Buckley et al 2006). This could possibly explain why these participants believed their child would cope. The ages of these participants’ children (average of 2 years old) were generally lower than those who believed their child would have to go to a special school (average age of 4 years old) and this may have also played a role in their perception of their child’s intellectual ability.

Seven out of the ten participants (P1, P2, P3, P6, P7 and P8) knew that DS was caused by a default in the genetic material, but very few knew what this default entailed. Only three (P6, P7 and P8) could recall that DS is caused by an extra chromosome 21. P4 had an idea that something was extra in the body, but could not say that it was a chromosome. The participant explained it in the following way:

*“As I’ve heard it is a sperm, something about the sperm that’s...was it now more in the body? In the man’s body or the woman’s body, one of the two, then it gets passed on and then it is how DS develops.” (Translated)*

The level of understanding regarding the causes of DS were not influenced by the type of DS their child had. All of the participants had a child with trisomy 21 type DS, except P1 who had a child with translocation type DS. However, P1 had a similar level of understanding as the others.

Overall the participants had a good general understanding of the features of DS and a basic understanding of the genetic cause of DS. As indicated in Figure 4.5 P7, P6 and P1 had the highest level of understanding and P9 and P2 had the lowest level of understanding.

It is uncertain what the reasons for the difference in understanding were between the participants as indicated in Figure 4.5 as it is not definite that income level and socio-economic status played a role. P1, P2 and P6 had high levels of income and socio-economic status, but their levels of understanding varied. It doesn’t seem that level of education played a role either as the participants who had the highest level of understanding had similar educational backgrounds as the one’s who had the lowest level. Also the two participants, P4 and P10, who had the lowest level of education, did not seem to understand less about DS than the other participants. These findings differ from that reported by other qualitative studies conducted by De Pina-Neto and Petean (1999) and Molster et al (2009). De Pina-Neto (1999) and Molster et al (2009) found that higher levels of education and income, as well as a higher socio-economic status, lead to higher levels of understanding in Brazilian and Australian individuals. In general it is difficult to make comparisons as the sample size included in the current study is much smaller than those reported in other studies.

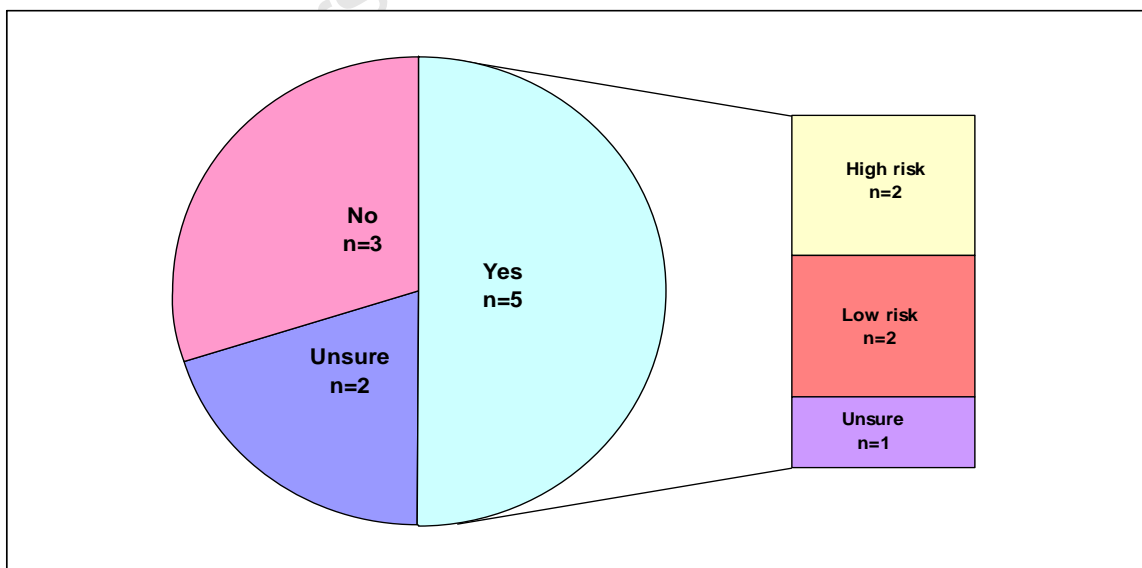
A factor that did seem to play a role in the participants’ understanding was their level of interaction with other individuals with DS. P6, P4 and P5 who had more experience with individuals with DS had a higher level of understanding than the other participants. This

finding is similar to that reported by Lucke et al (2008) and Walter et al (2004). Lucke et al investigated the knowledge of inherited colorectal cancer amongst a group of Australian individuals. Walter et al (2008) investigated the level of knowledge of the lay public in the UK regarding inherited common chronic diseases.

The genetic counselling the participants received also seemed to influence the participants' level of understanding. P2 had the lowest level of understanding which could possibly be explained by the fact that this participant had not received any counselling regarding the causes and features of DS. The only two participants (P1 and P2) who had received counselling from a genetic counsellor, had the highest level of understanding.

#### 4.8.2 Discussion of participants' knowledge of recurrence risks and preventive strategies

The participants were asked whether they thought it was possible to have another child with DS and what their understanding was regarding their risks. Figure 4.7 illustrates their responses.



**Figure 4.7: Participants' response to questions of whether possible to have future child with DS and participants' perceptions of risks (n=10)**

The knowledge of the participants regarding prenatal screening and testing options were also investigated. All of the participants knew about the possibility of prenatal diagnosis, but lacked the knowledge of screening tests. Seven (P2, P3, P4, P6, P7, P8 and P10) out of the ten participants had some idea of how the amniocentesis is performed. P1 knew that they took something out of the womb, but had the misconception that it was blood from the baby instead of the amniotic fluid. P6 was under the impression that the test was performed through the navel.

Out of the ten participants, three (P5, P7 and P10) were offered prenatal diagnosis by means of the amniocentesis procedure when they were pregnant with their child with DS. P5 decided not to have testing done due to their religious beliefs that you must *“accept what is coming your way.”* P7 and P10 both decided to have testing done, because they felt they needed to know whether the baby had DS to prepare themselves for the baby’s arrival.

The seven participants who were not offered any prenatal screening or diagnostic testing were asked whether they would have wanted the option. Two participants (P2 and P6) were ambiguous and felt that knowing had the benefit of preparing oneself before the arrival of the baby, but they also felt that it would cause them stress for the remainder of the pregnancy. Three participants (P3, P4 and P9) would not have wanted the option anyway, because they felt knowing before the birth of their child with DS would not have changed anything. Two participants (P1 and P10) felt that they would have wanted testing done and would have wanted to prepare themselves before birth rather than face the shock once the child was born.

### **4.8.3 Discussion of participants’ attitudes towards preventive strategies**

The participants were asked how they felt towards the preventive strategies that were offered to pregnant women to determine their risks of having a baby with DS. They were also asked what their attitudes were towards the option of terminating a pregnancy if the baby had DS. Their responses are shown in Table 4.9.



**Table 4.9: Participants attitudes towards preventive strategies**

P. No	Feelings regarding prenatal testing and screening	Feelings regarding TOP if foetus has DS
1	"I think they should do it with everybody...but then also it is your choice. So they can know before and give the mom the options to decide what to do."	"I don't know. I can't say. Everybody feels different. But then also it is your choice, because you are going to live with that conscience to kill your child while the child is living with you, because the moment there is a heartbeat the child is actually alive. So would you live with that conscience that you're killing your child, because it's a DS child? I don't think so."
2	"I think it is a good thing, because your pregnancy might be in danger, but then the doctors would have known."	"I would say that maybe they are just ungrateful, because they don't get to keep what the Man above has given them. Why terminate a pregnancy with a child with DS, because they are so special, what else would you have wanted?"
3	"I wouldn't go for it, because you read so much that it can harm your baby, you can miscarry, and I wouldn't want to do that to an unborn child. I wouldn't even recommend it to my best friends."	"No I wouldn't, I wouldn't abort. I will try and talk them (other mothers) out of it, but like I say that is their own decision."
4	"It is a good thing for other people, but not for me. I am a person that is content with whatever comes my way, but some younger mothers are very ungrateful." (translated)	"That's murder." (translated)
5	"I don't think it is a good thing, I won't encourage it, because whatever the Man from above gives us you must take. He won't give you something you can't handle."	"No matter what is wrong with the baby, don't stop the pregnancy. It's almost like I can tell my father: 'Listen don't you want me anymore then put a bag over my head and kill me.' It's the same thing."

6	"I think it is something good, because you get parents, like girls that are still very young. I think to know what problem you are getting is actually good, but I think it is not too good, because the wondering about if the child will make it or what the child will look like if he comes out is very traumatic." (translated)	"Everyone is not the same, I myself think every child deserves a life, whether it is a disabled child or whether it is what type of child, come let me give you a life. So I don't think it is our decision to take away that child. So if God, because He won't give you life if He didn't want you to have it, then He would've taken it away Himself. So I don't think abortion is something that should happen." (translated)
7	"I think it is a good thing. If they are interested and they want to know they should do it."	"They wanted to stop my pregnancy, but I said no, I want to keep the child, because I knew it was a Downs and I would recommend other mothers to do the same."
8	"I think it is a good thing, because you need to know about your child's health, because if you are not interested a lot of things will happen and it's rather best to know and to be prepared."	"I will never terminate, because it is really wrong. I mean after all that there is a life that you are carrying inside of you. It is an innocent baby that did nothing to anybody. It just wants to be in the world so I think its wrong for parents to abort, even before a month."
9	"A lot of people don't want to, but I think if you let the tests be done then you are prepared for what is waiting on you." (translated)	"As I understand I will recommend that you don't remove your child. If I had known (child with DS) had DS, I would not have removed her." (translated)
10	"For me it was a good idea, because if I had to give birth and they told me after that, I think it would have been more difficult to hear as when they told me before birth. I would recommend it to others." (translated)	"It is not right. For me it is if you are pregnant and it is a baby with DS, I don't know how others will feel, but for me it feels like you should go on with the pregnancy." (translated)

## CHAPTER 4: ANALYSIS, FINDINGS AND DISCUSSION

The majority of the participants felt positively about the prenatal screening and diagnostic testing options that were available. P3 and P5 were completely against these policies. P3 was concerned about the harm it could cause the unborn child and P5 was against it for religious reasons. These findings are similar to those reported by Lampret and Christianson (2007) and Pieters et al (2009) amongst SA mothers with a child with DS and pregnant women in the Netherlands respectively.

All of the participants were against the termination of a foetus that has DS. A possible explanation for this could be that because they have a child with DS, they feel that opting for a termination would be like symbolically killing this child that they already have. Another reason could be due to this group having an overall positive experience with having a child with DS and not seeing this child as a huge burden.

These findings differ from that reported by Lampret and Christianson (2007) who reported that slightly less than half of caucasian and black mothers with a child with DS would consider a TOP if the foetus had DS. This difference in views could possibly be attributed to the difference in ethnicity between the current study's population group and that of previous studies. This warrants further research.

## Chapter 5

### CONCLUSION



## 5.1 CONCLUSION

The aims of this study were to investigate the level of knowledge regarding DS and the needs of parents of mixed ancestry with a preschool child with DS. The study also aimed at exploring the extent to which these needs were met by the available health care and supportive services and to identify the problems these parents experienced in their everyday lives. All the findings of this study are novel and unique as no other reports have been made on how mixed ancestry parents in the WC experience having a child with DS.

Nine mothers and one father of a child with DS participated in the current study and their ages ranged between 19 years and 46 years old. The majority of the participants were unemployed, married and did not complete education on a secondary level. The participants were from the lower to middle income group and all of them lived in the lower socio-economic areas.

The majority of the participants were told about their child's diagnosis of DS postnatally and generally described this event as a shock. Even though the news itself was traumatic, the majority were satisfied with the way in which the health care professional had broken it to them. The findings suggest that religious beliefs and having past interactions with someone with DS played a protective role in the parents receiving this news.

This study once again highlighted the importance of breaking this news in a satisfactory manner. The two participants who felt the news delivery was unsatisfactory were traumatised by this event and still experience the emotions they felt at the time. This suggests the importance of properly training the health care staff in the labour wards on the appropriate manner in which the news should be broken and how important it is to use sensitive language.

The news of the diagnosis is not only a shocking experience for the parents of the child with DS, but also for the families. Even though the diagnosis was an unexpected event,

these families were accepting towards the child with DS and proved to be an irreplaceable source of support for the parents.

The majority of the participants were satisfied with the counselling they received and found the information invaluable. One participant appreciated the relief of guilt she experienced after receiving counselling. The participants who found the counselling lacking was counselled by health care professionals who were not appropriately trained in genetics. These participants felt that the information was not age appropriate and that the timing of the counselling was inappropriate. This indicates the importance of these parents receiving counselling by a genetic counsellor or medical geneticist as these professionals are trained to adequately address these parents' informational and psychosocial needs.

The parents were generally satisfied with the health care and supportive services they received from the RCWMCH and TBH. They felt these services adequately addressed their needs as well as the needs of their child with DS. Even though they were satisfied with the services itself they had some complaints about the clinics being overcrowded at the RCWMCH and the attitudes of the nursing staff at TBH.

All of the participants had to attend these hospitals on a regular basis as their children with DS had appointments at several specialised clinics. The majority of the participants struggled attending all their children's clinic appointments due to one or more of the following reasons: lack of time, transport problems, bad weather and the child being ill or hospitalised.

The majority of the participants were receiving an adequate amount of social support. This support was provided from various sources including their partners, family, friends, support groups and the church. The value of social support was highlighted as the only participant who was not receiving any form of support was having the most difficult time coping with her child with DS. Other factors that played an important role in the

parents' successful adaptation to their child with DS were support from their partner and adequate financial resources.

The participants mostly experienced having a child with DS positively. The majority felt that it had a positive effect on their marital relationship and their other children. The majority reported their children as being loving and helpful towards their sibling with DS. They reported that having a child with DS taught them to be compassionate and enriched their lives as well as the lives of the other family members with happiness and laughter.

As children with DS are more prone to infections and illnesses they are often hospitalised in their preschool years. The majority of the participants described their children's hospitalisations and illnesses as the most unpleasant aspect of about having a child with DS. The reason this is experienced negatively is because of the parents' constant fear of losing their child.

In general the participants had a good overall level of understanding of the features of DS. Knowledge about the facial features and developmental delay of these children were the best understood. The areas where knowledge was lacking was regarding the other physical features of DS and their intellectual ability. Only half of the participants acknowledged that children with DS have an intellectual disability. Half of the parents believed that their child would be able to cope in a mainstream school; however this finding was mostly found in the parents who have younger children with DS. The majority of the participants knew that DS was caused by an anomaly in the genetic material, but could not recall that this anomaly was an extra chromosome 21. The participants' level of education, financial status and socio-economic status did not seem to influence their understanding of DS. Factors that seemed to increase the level of understanding was increased personal experience and interaction with individuals with DS, as well as having received counselling from a genetic counsellor.

Half of the participants were aware of the risk of having another baby with DS. The participants had good knowledge of the available prenatal testing options for future pregnancies, but lacked knowledge on the available screening tests. The majority had a positive attitude towards prenatal testing; however all of the participants were against the termination of a pregnancy if the foetus had DS.

The parents attending the RCWMCH and TBH felt that these services were meeting their needs and did not feel there was much room for improvement. The information regarding the experiences of these parents and their daily struggles are important for health care professionals working with mixed ancestry parents with a child with DS. This insight will undoubtedly lead to greater sensitivity to their situation and help promote their adaptation to their child with DS. Parents' perception of their child with DS and how they cope with him/her is important information to take into consideration when counselling parents with a newly diagnosed child with DS. This information-giving process should continue throughout the life of the child and should not only take place shortly after the diagnosis has been made. Hopefully with an increased awareness amongst health care professionals there could be a vast improvement in the ability of these families to cope.

“With our sensitive appreciation of the grief process, the clients' chaotic feelings can be transformed into positive behaviour. Thus the grief can become a sadness that enables the clients to appreciate what they have, the anger can become the energy to make the change, the guilt can become the commitment, the recognition of vulnerability can become the means by which clients reorder priorities, and the resolution of confusion can become the motivation for learning” (Luttherman 1996:62).



## Chapter 6

### RECOMMENDATIONS



## 6.1 RECOMMENDATIONS

Based on the outcomes of the study, the following recommendations are made:

- Information or training courses on how to communicate the news of the diagnosis of DS to families in a better way offered to the obstetricians, paediatricians and nursing sisters at the various day hospitals. As the health care professionals at the day hospital are usually the first ones to break this news to the parents, this information could be of benefit to try and make this event as positive for the parents as possible. Hospital staff could benefit from increased awareness regarding DS.
- Health care professionals breaking the news may make this experience less traumatic by taking the following recommendations made by Skotko et al (2009) into consideration. Parents seem to find this experience less traumatic if they are informed of the suspicions of the doctors even before the diagnosis is confirmed. It is recommended that the parents are taken to a private room away from the other patients and visitors when they are told. It is advisable to inform the parents together with their infant with DS present. They should congratulate the parents and focus more on the positive things regarding DS. They should provide the parents with accurate and up-to-date information on DS.
- Health care professionals should be attentive to the words they use when speaking to the parents about their child with DS. They should be sensitive towards the parents' feelings and use compassionate language.
- Health care professionals should provide families with information and contact details regarding their local parent support groups. These support groups have demonstrated to be an important source of support for parents who lack social support from other resources.
- Genetic Counselling should be aimed at limiting blame. It is recommended that information regarding the causes of DS should be given in a neutral way and should emphasise that it is uncertain who contributed the extra chromosome 21 to the child. Emphasis that it is no one's fault limits parents' feelings of guilt and promotes their coping process.

- Age appropriate information should be given to the parents during the initial counselling session. It is important to give information according to the age of the child at that stage as parents' find it more applicable to their situation.
- Information-giving should not stop at the initial session when the diagnosis is given. Parents of children with DS should be offered follow-up appointments with a genetic counsellor or clinical geneticist to discuss any questions they might have regarding their child. These appointments should take place at different stages of their child's development as they might experience different informational needs at different points in time.
- Management should be based on family-centred care. The support these parents receive from their families is crucial for their successful adaptation to their child with DS. Therefore it is recommended that the management options offered to these parents should be focused on the care of the whole family and that they should be included in decisions about the child's future management.
- Better education of women, especially pregnant women, regarding the risks of having a child with DS and the prenatal screening and diagnostic testing options. This might help motivate mothers to initiate antenatal care at an earlier stage to accurately assess their risks.

### **6.2 RECOMMENDATIONS FOR FUTURE RESEARCH**

It will be of great value to:

- Conduct a similar study with a larger population group. The current study included a very small number of parents with a child with DS and it is therefore important to do a study on a larger group to validate the findings.
- Investigate the reasons why parents have never attended any of the health care and supportive services.
- Investigate the experiences of parents in other ethnic groups or geographical areas in SA as the current study is only representative of a small group of the mixed ancestry population in the Cape Town area.

- Investigate the knowledge of DS amongst health care professionals working at various delivery wards and how comfortable they are delivering a diagnosis of DS to parents.
- Investigate the experience of fathers regarding having a child with DS. In the current study only one father was interviewed and it would be interesting to do interviews with only the fathers and compare it to the views of the mothers or how the mothers perceive their partners' experiences.
- Further investigation regarding the positive aspects of having a child with DS. To provide families with the correct information on DS it is important to focus on the positive aspects and not only the negative aspects. Knowledge regarding the positive aspects is limited as many research studies have only focused on the negative aspects.
- Investigate the knowledge of the siblings of a child with DS and how they experience living with their sibling. The current study provided information on how the parents' perceived the attitudes of the siblings and it would be valuable to compare this with the actual feelings experienced by the siblings.

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## APPENDIX I

**ENGLISH VERSION OF INFORMATION AND CONSENT FORM**

MSc in Genetic Counselling Research Project

**An investigation into the experiences of parents with  
a preschool child with Down syndrome**

**INFORMATION AND CONSENT FORM**

*STATEMENT BY PARTICIPANT*

I, ....., living at  
(address).....  
confirm that:

1. I have been invited to participate in the above research project which has been initiated through the Division of Human Genetics, University of Cape Town because I have a preschool child who has Down syndrome (DS) and attend the clinics and supportive services rendered at the Red Cross War Memorial Children's Hospital (RCWMCH) or Tygerberg Hospital (TBH).

2.1. I understand that the objectives of this study is to investigate:

- the level of knowledge of DS;
- the experiences of the news being broken regarding our child's diagnosis;
- the impact of our child's diagnosis on ourselves and our families;
- why we do/don't regularly attend follow-up sessions with health care professionals; and
- the level of satisfaction we experience with the services at RCWMCH or TBH.

- 2.2. I understand that the interview will take place in my home or another venue of my choice and that it may take one or two visits of two hours each.
- 2.3. I am aware that this is a once-off investigation that will take place in 2010 at a time convenient to me and my family.
- 2.4. I understand that some of the questions may make me angry or sad, but the risks from the study are minimal. The researcher will refer me to a genetic counsellor if necessary. She will show me respect, acceptance and empathy during the interview.
- 3.1. I have been assured that all information will be handled confidentially. Information may be used for a thesis, publication in scientific journals and presentations at professional congresses, but names will not be included.
- 3.2. I understand that the interview will be audio-recorded so that the researcher does not have to write too much during the interview. The recordings will be stored in a safe until the research has been written up and will then be destroyed immediately. The recordings and the data stored on the computer will have a numerical code only and my name does not appear anywhere.
4. I have been assured that the recorded and transcribed information discussed at the meeting will only be made available to the researcher's supervisors with my study code number and that they do not know that it refers to my name.
5. I have not been coerced to consent to taking part in the study and I have been informed that I may refuse to participate in this project and that I may stop participating at any stage, and that such refusal or stoppage will not negatively affect my current or future access to medical and genetic services to which I am entitled.



6. .... has explained the information of the study to me in English/Afrikaans. I am proficient in that language and my questions have been answered satisfactorily.
7. I understand that there will be no medical benefits to me from this study.
8. I have been assured that participation in this project will not lead to additional costs for me or my family and I will not benefit from it financially.

**I HEREBY DECLARE THAT I VOLUNTARILY AGREE TO PARTICIPATE IN THE ABOVE RESEARCH STUDY**

Signed at: (address)..... on  
.....2010

Participant's signature

Witness signature

**I HEREBY DECLARE THAT I AGREE TO HAVE MY INTERVIEW AUDIO-RECORDED**

Signed at: (address).....on  
.....2010

Participant's signature

Witness signature

**IMPORTANT INFORMATION**

Dear Participant,

Thank you for your participation in this study. Should you have any questions during the duration of this study regarding:

1. problems as a result of the research, or
2. questions regarding information about the project

please feel free to contact me at the following telephone number:

Chantelle Scott: (021) 406 6373

Email: [chantelle.scott@uct.ac.za](mailto:chantelle.scott@uct.ac.za)

Prof. Jacquie Greenberg: (021) 406 6299

If you have any questions regarding your right as a participant, contact Prof. Marc Blockman, the chairman of the research ethics committee of the Faculty of Health Sciences, University of Cape Town on (021) 406 6492.

**AFRIKAANS VERSION OF INFORMATION AND CONSENT FORM**

MSc in Genetiese Raadgewing Navorsingsprojek

**‘n Ondersoek na die ervarings van ouers met ‘n voorskoolse kind met  
Down sindroom****INLIGTING EN TOESTEMING VORM***VERKLARING DEUR DEELNEMER*

Ek, ....., wat woon by  
(adres) .....

bevestig dat:

1. Ek uitgenooi is om aan die bogenoemde navorsingsprojek deel te neem wat geïnisieer is deur die Divisie van Mensgenetika, Universiteit van Kaapstad, aangesien ek ‘n voorskoolse kind het met Down sindroom (DS) en die klinieke en ondersteunende dienste wat aangebied word by die Rooi Kruis Oorloggedenk Kinderhospitaal (RKOGKH) of by die Tygerberg hospital (TBH), bywoon.

2.1. Ek verstaan dat die doel van hierdie projek is om die volgende te ondersoek:

- kennis van DS;
- die ervaring van hoe die nuus oorgedra is dat ons kind DS het;
- die impak van ons kind se diagnose op onself en ons familie;
- die rede hoekom ons/hoekom ons nie gereeld ons opvolg afsprake by die klinieke bywoon; en
- die vlak van tevredenheid wat ons ondervind met die dienste by RKOGKH or TBH.

2.2. Ek verstaan dat die onderhoud of by my huis of by ‘n ander plek van my keuse sal plaasvind en dat dit een of twee besoeke van twee ure elk behels.

- 2.3. Ek is bewus dat dit 'n eenmalige onderhoud is wat in 2010 sal plaasvind op 'n tyd wat vir my en my gesin gerieflik is.
- 2.4. Ek verstaan dat sommige van die vrae my hartseer of ongelukkig mag laat voel, maar dat die risiko's van die studie minimaal is. Die navorser sal my na 'n genetiese raadgewer verwys indien nodig. Sy sal my met respek, aanvaarding en empatie behandel gedurende die onderhoud.
- 3.1. Ek is verseker dat alle inligting vertroulik behandel sal word. Inligting mag vir 'n tesis, publikasie in wetenskaplike joernale en aanbiedings by professionele kongresse gebruik word, maar name sal nie ingesluit word nie.
- 3.2. Ek verstaan dat die onderhoud opgeneem sal word sodat die navorser nie te veel hoef te skryf gedurende die onderhoud nie. Die opname sal in die kluis gestoor word tot die navorsing opgeskryf is en sal daarna dadelik vernietig word. Die opname en data op die rekenaar sal slegs 'n numeriese kode op hê en my naam sal nie daarop verskyn nie.
4. Ek is verseker dat die inligting wat opgeneem en getranskibeer is slegs aan die navorser se studie-leier bekend gemaak word, maar dit sal slegs my numeriese studie kode bevat en my naam sal nie daarop verskyn nie.
5. Ek neem vrywilliglik deel aan die projek en ek is bewus dat ek mag weier om deel te neem, en ek kan op enige stadium besluit om te onttrek. My onttrekking sal op geen manier my huidige of toekomstige toegang tot die mediese of genetiese dienste, waarop ek geregtig is beïnvloed nie.
6. .... het die inligting van die projek aan my verduidelik in Engels/Afrikaans. Ek is vlot in hierdie taal en my vrae is ten volle beantwoord.

7. Ek verstaan dat daar geen mediese voordele vir my sal wees as gevolg van hierdie projek nie.
8. Ek is verseker dat my deelname in hierdie projek nie tot enige addisionele koste vir my en my families sal lei nie en dat ek nie finansieel daarby gaan baat nie.

**EK VERKLAAR HIERMEE DAT EK VRYWILLIGLIK AAN DIE BOGENOEMDE NAVORSINGSPROJEK DEELNEEM**

Geteken te: (adres)..... op  
.....2010

.....  
Handtekening van Deelnemer

.....  
Handtekening van Getuie

**EK VERKLAAR HIERMEE DAT EK TOESTEMING GEE DAT MY ONDERHOUD OPGENEEM MAG WORD**

Geteken te: (adres)..... op  
.....2010

.....  
Handtekening van Deelnemer

.....  
Handtekening van Getuie

**BELANGRIKE INLIGTING**

Geagte Deelnemer,

Baie dankie vir u deelname aan hierdie studie. As u gedurende die verloop van die navorsing enige vrae het aangaande:

1. probleme as gevolg van die navorsing, of
2. vrae aangaande die inligting oor die projek

kontak my of Prof. Greenberg gerus op die volgende telefoonnommers :

Chantelle Scott: (021) 406 6373

Epos: [chantelle.scott@uct.ac.za](mailto:chantelle.scott@uct.ac.za)

Prof. Jacquie Greenberg: (021) 406 6299

As u enige vrae het in verband met u regte as 'n deelnemer, kontak Prof. Marc Blockman, die Voorsitter van die Navorsings Etiek Hersienings Komitee van die Fakulteit van Gesondheidswetenskappe, Universiteit van Kaapstad by (021) 406 6429.

## APPENDIX II

**CONSENT FORM FOR USE OF PHOTOGRAPHS**

UNIVERSITY OF CAPE TOWN

Division of Human Genetics

**CONSENT FOR PHOTOGRAPHS**

My name is ..... I have been informed that the staff of the  
UCT Division of Human Genetics would like to obtain photographs of my child

.....

I have been informed that the photographs will only be used for specific purposes for  
which I give consent, as follows (please initial or tick):

- ☐ Publication in a dissertation/thesis (no names will be included)
- ☐ Presentation at a medical/scientific conference or seminar (no names will be included)
- ☐ Publication in a medical/scientific journal (no names will be included)

Signed: .....

Date: .....

Informant/witness signature: .....

Date: .....

## APPENDIX III

**ENGLISH VERSION OF INTERVIEW SCHEDULE**

Participant Code Number:.....

**A. Sociodemographic Information**

## 1. Family history

- Date of birth of both parents (if available)?
- Age of mother when pregnant with child with Down syndrome (DS)?
- Marital status?
- How many children?
- Ages of children?
- Are the children all well?
- Age of child with DS?
- Is child with DS first/second/third/fourth born?

## 2. Where do you live? (area)

## 3. What type of housing do you have?

- |  |  |
|--|--|
| <input type="checkbox"/> Wendy house             | <input type="checkbox"/> House         |
| <input type="checkbox"/> Flat                    | <input type="checkbox"/> Room in house |
| <input type="checkbox"/> Shack or informal house |  |

## 4. How many rooms are there in your home?

- |                                   |                                  |
|-----------------------------------|----------------------------------|
| <input type="checkbox"/> Bedroom  | <input type="checkbox"/> Kitchen |
| <input type="checkbox"/> Bathroom | <input type="checkbox"/> Other:  |



5. How many people live in the home with you?
6. What is their relationship to you?
- If not family, how are they related?
7. Which grade/standard did you complete at school? (both parents)
- |   |  |
|---|--|
| <input type="checkbox"/> Grade 12<br>(matric) | <input type="checkbox"/> Grade 9 (Std 7) |
| <input type="checkbox"/> Grade 11<br>(Std 9)  | <input type="checkbox"/> Grade 8 (Std 6) |
| <input type="checkbox"/> Grade 10<br>(Std 8)  | <input type="checkbox"/> Grade 7 (Std 5) |
|   | <input type="checkbox"/> Grade 6 (Std 4) |
|   | <input type="checkbox"/> Other:          |
8. Have you started any further courses/training since leaving school?
- |                              |                             |
|------------------------------|-----------------------------|
| <input type="checkbox"/> Yes | <input type="checkbox"/> No |
|------------------------------|-----------------------------|
9. If Yes to question 8, what?
- |  |
|--|
| <input type="checkbox"/> Trade/Apprentice            |
| <input type="checkbox"/> Certificate from college    |
| <input type="checkbox"/> Diploma                     |
| <input type="checkbox"/> Bachelor's degree           |
| <input type="checkbox"/> Postgraduate diploma/degree |
| <input type="checkbox"/> In service training         |
| <input type="checkbox"/> Other                       |
10. Have you completed it? If not, give reasons.
- |                              |                             |
|------------------------------|-----------------------------|
| <input type="checkbox"/> Yes | <input type="checkbox"/> No |
|------------------------------|-----------------------------|
11. Are you currently working in a permanent full-time job? (both parents)
- |                              |                             |
|------------------------------|-----------------------------|
| <input type="checkbox"/> Yes | <input type="checkbox"/> No |
|------------------------------|-----------------------------|

12. If No to question 11, are you?

- ☐ Unemployed
- ☐ Unable to work due to caring for child with DS
- ☐ Housewife
- ☐ Full-time student
- ☐ Part-time student
- ☐ Unfit for work
- ☐ Retired/Pensioner
- ☐ Casual employment
- ☐ Other:

13. Is the work...?

- ☐ Self-employed permanent/temporary
- ☐ Full-time permanent/temporary
- ☐ Part-time permanent/temporary
- ☐ Other:

14. How many people contribute to the household income?

15. How many people does the household income support?

16. What is the current household income per month?

- ☐ No income
- ☐ Disability or child support grant
  - Number of people receiving these grants
- ☐ Salary income
  - R1 – R500
  - R501 – R1 000
  - R1 001 – R1 500
  - R1 501 – R2 000

- R2 001 – R5 000
- R5 001 – R10 000
- R10 001 – R15 000
- R15 001 – R30 000
- R30 001 – R50 000
- R50 001 – R100 000
- R100 001 – R200 000
- More than R200 000

### **B. Breaking the News**

17. When did you first suspect a problem with your child?

- Can you describe the early signs that made you wonder if your child had a problem?

18. How old was your child when he/she was diagnosed with DS?

19. Who first told you that your baby had DS?

20. Describe under which circumstances you were told about your child's diagnosis?

- Where were you told about your child's diagnosis?
- Was it in quiet environment/ room?
- Were there any other people in the room with you?

21. Describe your experience of this event?

22. Describe the manner in which you would have liked to be told about your child's diagnosis?

- How do you think the news could have been broken to you in a better way?

23. Did you share the news of your child's diagnosis with anyone?

☐ Yes

☐ No

24. In No to question 23, describe reasons for not sharing the diagnosis?

- Does your family currently know that there is something different about your child?

25. If Yes to question 23, with whom did you share the diagnosis?

26. Describe how they reacted when you told them the news of your child's diagnosis?

27. Describe how you felt and experienced having to tell other people about your child's diagnosis?

28. Describe what this time was like for your family?

- How did your family find out or hear about your child's diagnosis?

### C. Satisfaction with Counselling Service

29. Did you receive any counselling regarding the cause of DS and its prognosis?

☐ Yes

☐ No

30. If Yes to question 29, from whom did you receive counselling?

☐ Genetic

☐ Paediatrician

counsellor

☐ Clinical geneticist

☐ Nurse/Sister

☐ Other:

31. When were you counselled?

32. Where did the counselling take place?

33. Describe how you experienced the counselling you received?

34. Do you feel the counsellor addressed your needs adequately?  
(informational/psychosocial)

☐ Yes

☐ No

35. If Yes to question 34, what did you experience positively about the counselling you received?

- What did you find most useful about the counselling?
- What did you like the most about the counselling?

36. If No to question 34, how would you have liked the counselling to be different to address your needs more adequately?

**D. Health Care Services at the Red Cross War Memorial Children's Hospital (RCWMCH) or Tygerberg Hospital (TBH)**

37. To which hospital do you take you your child with DS?

☐ RCWMCH

☐ TBH

☐ Other:

38. How far is it from your house?

39. How do you get to the hospital?

☐ Own transport

☐ Taxi

☐ Bus

☐ Other:

☐ Train

40. How much does it cost to get to the hospital?

41. How often do you and your child attend the health care services at the hospital?

<b>Clinic/ Service</b>	<b>Often</b>	<b>Not often</b>
Developmental		
Speech		
Genetic		
Genetic Counselling		
Social Worker		
Paediatrician		
Toy Library		

42. If not often: What are your reasons for not attending these clinics on a regular basis?

43. Do you feel the health services at the hospitals are suitable for your child with DS?

- Do you feel that it is helpful for you and your child?
- Do you feel that it is beneficial to attend the services?

44. Do you currently have health concerns about your child which you feel are not being adequately addressed?

☐ Yes

☐ No

45. If Yes to question 44, what are your health concerns?

46. Describe your experience of the health care services offered to your child with DS?

47. What recommendations do you have, if any, on how to improve the health care services?

**E. Social Support**

48. Who is providing you with the most support regarding your child?
- Who helps you the most with your child with DS? (Bathing, dressing, hospital visits)
49. What is your experience regarding support from family members and your partners?
50. Do you feel that you are receiving enough social support?
- ☐ Yes ☐ No
51. If No to question 50, what other supports would you like?
52. When you seek help with your child's health care problems, who helps you and how helpful are they?

**F. Experiences of Parents of Preschool Child with DS**

53. In general, what do you experience as a major problem in your daily lives?
- What do you struggle with in your daily life?
54. What are the emotional problems that you experience?
55. Has your child's diagnosis had an effect on your marital relationship?
56. How have your other children reacted towards their sibling with DS?
57. How would you describe the personality of your child with DS?
58. Describe how you have experienced the behaviour of your child with DS?
59. What has been the most unpleasant experience for you?

60. How would you have wanted this unpleasant experience to be different?

- How would you want to change this experience?

61. Is there a positive experience with having a child with DS?

- What is pleasant about having a child with DS?
- What do you like about having a child with DS?

### **G. Level of Understanding of the Genetics and Features of DS**

62. What is your understanding about DS and the problems children with this condition have?

- How are children with DS different from other children?
- Do you think your child will be able to cope in a mainstream school?

<b>Characteristic</b>	<b>Specific response</b>	<b>Participant score</b>	<b>Maximum score</b>
Facial features	Eyes, flat nasal bridge, protruding tongue		3
Physical features	Hypotonia, hands, sandal gap		3
Development	Delayed walking, delayed speech		2
Intellectual ability	Acknowledge intellectual disability		1
Heart problems	Acknowledge heart problem		1
Cause	Genetic/mistake in inherited material, extra chromosome/inherited material		2

63. Is it possible to have another baby with DS?

- ☐ Yes  
☐ No



64. If Yes to question 63, what is your understanding about your chances of having another baby with DS?

65. Is it possible to determine whether a baby has DS before birth?

- ☐ Yes
- ☐ No

66. If Yes to question 65, explain your understanding of how this is possible?

67. Were you offered any prenatal testing or screening when you were pregnant with your child with DS?

- ☐ Yes
- ☐ No

68. If Yes to question 67, what testing/screening were you offered?

69. Did you have any of the testing/screening done to determine your risk to have a baby with DS?

- ☐ Yes
- ☐ No

70. Describe your feelings regarding prenatal testing and screening?

- Would you recommend it to other people?

71. Describe your feelings regarding the termination of pregnancy if the foetus has DS?

**AFRIKAANS VERSION OF INTERVIEW SCHEDULE****Deelnemer Kode Nommer:.....****A. Sosiodemografiese Inligting**

## 1. Familie geskiedenis

- Geboortedatum van beide ouers (indien beskikbaar)?
- Ouderdom van moeder tydens swangerskap van kind met Down sindroom (DS)?
- Huwelikstatus?
- Hoeveel kinders?
- Ouderdomme van kinders?
- Is die kinders almal gesond?
- Ouderdom van kind met DS?
- Is kind met DS eerste/tweede/derde/vierde kind?

## 2. Waar woon u? (area)

## 3. Watter tipe behuising het u?

- |                                       |   |
|---------------------------------------|---|
| <input type="checkbox"/> Wendy huis   | <input type="checkbox"/> Huis             |
| <input type="checkbox"/> Woonstel     | <input type="checkbox"/> Kamer binne huis |
| <input type="checkbox"/> Plakkershuis |   |

## 4. Hoeveel kamers is daar in u huis?

- |                                     |                                  |
|-------------------------------------|----------------------------------|
| <input type="checkbox"/> Slaapkamer | <input type="checkbox"/> Kombuis |
| <input type="checkbox"/> Badkamer   | <input type="checkbox"/> Ander:  |

## 5. Hoveel mense woon saam met u in die huis?

6. Hoe is hulle verwant aan u?

- Indien nie familie, wat is hulle van u?

7. Watter graad/standard het u op skool voltooi? (beide ouers)

- |  |  |
|--|--|
| <input type="checkbox"/> Graad 12<br>(matriek) | <input type="checkbox"/> Graad 9 (Std 7) |
| <input type="checkbox"/> Graad 11<br>(Std 9)   | <input type="checkbox"/> Graad 8 (Std 6) |
| <input type="checkbox"/> Graad 10<br>(Std 8)   | <input type="checkbox"/> Graad 7 (Std 5) |
|  | <input type="checkbox"/> Graad 6 (Std 4) |
|  | <input type="checkbox"/> Ander:          |

8. Het u enige verdere kursesse/opleiding begin vandat u skool verlaat het?

- |                             |                              |
|-----------------------------|------------------------------|
| <input type="checkbox"/> Ja | <input type="checkbox"/> Nee |
|-----------------------------|------------------------------|

9. Indien Ja op vraag 8, wat?

- |   |   |
|---|---|
| <input type="checkbox"/> Ambag                      | <input type="checkbox"/> Baccalureurs graad         |
| <input type="checkbox"/> Sertifikaat<br>van kollege | <input type="checkbox"/> Nagraadse<br>diploma/graad |
| <input type="checkbox"/> Diploma                    | <input type="checkbox"/> In diens opleiding         |
|   | <input type="checkbox"/> Ander:                     |

10. Het u dit voltooi? Indien nie, gee redes.

- |                             |                              |
|-----------------------------|------------------------------|
| <input type="checkbox"/> Ja | <input type="checkbox"/> Nee |
|-----------------------------|------------------------------|

11. Het u huidiglik 'n permanente voltydse werk? (beide ouers)

- |                             |                              |
|-----------------------------|------------------------------|
| <input type="checkbox"/> Ja | <input type="checkbox"/> Nee |
|-----------------------------|------------------------------|

12. Indien Nee op vraag 11, is u?

- ☐ Werkloos
- ☐ Nie moontlik om te werk as gevolg van sorg vir kind met DS
- ☐ Huisvrou

- ☐ Voltydse student
- ☐ Deeltydse student
- ☐ Ongeskik vir werk
- ☐ Afgetree/ pensionaris
- ☐ Casual
- ☐ Ander:

13. Is die werk...

- ☐ U eie besigheid
- ☐ Voltyds permanent/tydelik
- ☐ Deeltyds permanent/tydelik
- ☐ Ander:

14. Hoeveel mense dra by tot die huishoudelike inkomste?

15. Hoeveel mense word onderhou deur die huishoudelike inkomste?

16. Wat is die huidige huishoudelike inkomste per maand?

- ☐ Geen inkomste
- ☐ Ongeskikheidstoelaag
  - Aantal mense wat toelaag ontvang:
- ☐ Salaris inkomste
  - R1 – R500
  - R501 – R1 000
  - R1 001 – R1 500
  - R1 501 – R2 000
  - R2 001 – R5 000
  - R5 001 – R10 000
  - R10 001 – R15 000
  - R15 001 – R30 000
  - R30 001 – R50 000

- R50 001 – R100 000
- R100 001 – R200 000
- Meer as R200 000

## **B. Oordra van die Nuus**

17. Wanner het u vir die eerste keer vermoed dat u kind 'n probleem het?

- Kan u vir my beskryf wat die vroeë tekens was wat u laat wonder het of u kind 'n problem het?

18. Hoe oud was u kind toe hy/sy gediagnoseer was met DS?

19. Wie het eerste vir u gesê dat u baba DS het?

20. Beskryf onder watter omstandighede u vertel was van u kind se diagnose?

- Waar was u vertel van u kind se diagnose?
- Was dit in stil omgewing?
- Was daar ander mense saam met u in die kamer?

21. Beskryf hoe u hierdie gebeurtenis ondervind het?

22. Beskryf die manier waarop u eerder wou gehad het die nuus aan u oorgedra moes word?

- Hoe dink u kon die nuus op 'n beter manier aan u oorgedra geword het?

23. Het u die nuus van u kind se diagnose met enige iemand gedeel?

☐ Ja

☐ Nee

24. Indien Nee op vraag 23, beskryf die redes hoekom u nie die nuus gedeel het nie?

- Weet u familie en vriende huidiglik dat daar iets anders is van u kind?

25. Indien Ja op vraag 23, met wie het u die nuus gedeel?

26. Beskryf hoe hulle gereageer het toe u hulle vertel het van u kind se diagnose?

27. Beskryf hoe u gevoel het en hoe u dit ervaar het om vir ander mense te vertel van u kind se diagnose?

28. Beskryf hoe u gesin hierdie tydperk ervaar het?

- Hoe het u gesin uitgevind of gehoor van u kind se diagnose?

### C. Tevredenheid met Beradingsdienste

29. Het u enige berading of inligting ontvang ten opsigte van die oorsake van DS en u kind se toekoms?

☐ Ja

☐ Nee

30. Indien Ja op vraag 29, van wie het u berading ontvang?

☐ Genetiese  
raadgewer

☐ Paediater

☐ Genetiese Dokter

☐ Sister

☐ Ander:

31. Wanneer het u berading ontvang?

32. Waar het berading plaasgevind?

33. Beskryf hoe u die berading wat u ontvang het ervaar het?

34. Voel u die berader het ten volle na u behoeftes omgesien?  
(inligtings/psigososiale)

☐ Ja

☐ Nee

35. Indien Ja op vraag 34, wat was vir u 'n positiewe ervaring van die berading wat u ontvang het?

- Wat was vir u die beste deel van die berading?
- Wat het vir u die meeste gehelp/beteken?

36. Indien Nee op vraag 34, watter behoeftes moes die berader meer deeglik na omgesien het?

**C. Gesondheidsdienste by die Rooi Kruis Oorloggedenk Kinder Hospitaal (RKOGKH) of Tygerberg Hospitaal (TBH)**

37. Na watter hospitaal neem u u kind met DS?

- |                                 |                              |
|---------------------------------|------------------------------|
| <input type="checkbox"/> RKOGKH | <input type="checkbox"/> TBH |
| <input type="checkbox"/> Ander: |                              |

38. Hoe ver is dit van u huis af?

39. Hoe kom u by die hospitaal?

- |                                      |                                 |
|--------------------------------------|---------------------------------|
| <input type="checkbox"/> Eie vervoer | <input type="checkbox"/> Taxi   |
| <input type="checkbox"/> Bus         | <input type="checkbox"/> Ander: |
| <input type="checkbox"/> Trein       |                                 |

40. Hoeveel kos dit om by die hospitaal uit te kom?

41. Hoe gereeld woon u en u kind die gesondheidsdienste by die hospitaal by?

Kliniek/ Diens	Gereeld	Nie gereeld
Developmental		
Spraak		
Genetika		
Genetiese Raadgewing		
Maatskaplike werker		
Paediater		
Toy Library		

42. Indien nie gereeld: Wat is die redes hoekom u nie gereeld hierdie klinieke bywoon nie?

43. Voel u die gesondheidsdienste by die hospitale is geskik vir u kind met DS?

- Voel u dat dit uself en u kind help?
- Voel u dat dit voordelig is om die dienste by te woon?

44. Het u huidiglik enige gesondheidsbekkommernisse oor u kind wat u voel nie ten volle na omgesien word nie?

☐ Ja

☐ Nee

45. Indien Ja op vraag 44, wat is u gesondheidsbekkommernisse?

46. Beskryf u ervaring van die gesondheidsdienste wat aangebied word vir u kind met DS?

47. Watter voorstelle het u, indien enige, oor hoe om die gesondheidsdienste te verbeter?



**D. Sosiale Ondersteuning**

48. Oor die algemeen, wie ondersteun u die meeste ten opsigte van u kind met DS?

- Wie help u die meeste met u kind met DS? (Bad, aantrek, hospitaal besoeke)

49. Hoe ervaar u die ondersteuning van u familie en lewensmaat?

50. Voel u dat u genoeg sosiale ondersteuning ontvang?

☐ Ja

☐ Nee

51. Indien Nee op vraag 50, watter ander ondersteuning het u nodig?

52. Wanneer u help nodig het met u kind se gesondheids probleme, wie help vir u en hoe hulpvaardig is hulle?

**E. Ervaring van Ouers van Voorskoolse Kinders met DS**

53. Oor die algemeen, wat ervaar u as die grootste problem in u alledaagse lewe?

- Wat is vir u die moeilikste in u alledaagse lewe?
- Waarmee sukkel u elke dag?

54. Watter emosionele probleme ondervind u?

55. Het u kind se diagnose 'n effek gehad op u huwelik? Indien ja, beskryf wat die effek was.

56. Hoe reageer u ander kinders teenoor hulle broer/suster met DS?

57. Hoe sal u die persoonlikheid van u kind met DS beskryf?

58. Beskryf hoe u die gedrag van u kind met DS ervaar?

59. Wat was die mees onaangename ervaring wat u gehad het?

60. Hoe sou u hierdie onaangename ervaring anders wou gehad het?

- Hoe sou u hierdie ervaring wou verander?

61. Is daar 'n positiewe ervaring om 'n kind te hê met DS?

- Wat is vir u 'n goeie ding/lekker daarvan om 'n kind met DS te hê?

#### **F. Vlak van verstaan van die genetika en kenmerke van DS**

62. Wat verstaan u rondom DS en die probleme wat kinders met hierdie kondisie ondervind?

- Hoe is kinders met DS anders as ander kinders?
- Dink jy u kind sal kan aanpas in 'n hoofstroom/gewone skool?

<b>Eienskap</b>	<b>Spesifieke eienskappe</b>	<b>Deelnemer telling</b>	<b>Maksimum telling</b>
Gesigskenmerke	Oë, plat brug van neus, tong		3
Fisiese kenmerke	Lae spiertonus, hande, gaping tussen groottoon en tweede toon		3
Ontwikkeling	Vat langer om te stap, praat eers op later stadium		2
Intellektuele vermoë	Erken intellektuele gestremdheid		1
Hart probleme	Erken dat hart probleem algemeen is		1
Oorsake	Genetiese probleem/ probleem in oorerflike materiaal, ekstra oorerflike materiaal/chromosoom		2

63. Is dit moontlik om nog 'n baba met DS te kry?

- ☐ Ja  
☐ Nee

64. Indien Ja op vraag 63, wat verstaan u rondom u kanse om nog 'n baba te kry met DS?

65. Is dit moontlik om te bepaal of 'n baba DS het voorgeboorte?

- ☐ Ja
- ☐ Nee

66. Indien Ja op vraag 65, verduidelik wat u verstaan rondom hierdie moontlikheid?

67. Was u enige voorgeboorte toetsing aangebied terwyl u swanger was met u kind met DS?

- ☐ Ja
- ☐ Nee

68. Indien Ja op vraag 67, watter toetse was u aangebied?

69. Het u enige toetsing laat doen om te bepaal wat u risiko was om 'n baba te kry met DS?

- ☐ Ja
- ☐ Nee

70. Beskryf u gevoelens rondom voorgeboorte toetsing?

- Sal u dit vir ander mense aanbeveel?

71. Beskryf u gevoelens rondom die stop van 'n swangerskap indien die fetus DS het?

## APPENDIX IV

**LETTER OF ETHICAL APPROVAL**

UNIVERSITY OF CAPE TOWN



Health Sciences Faculty  
 Research Ethics Committee  
 Room E52-24 Groote Schuur Hospital Old Main Building  
 Observatory 7925  
 Telephone [021] 406 6626 • Facsimile [021] 406 6411  
 e-mail: shuretra.thomas@uct.ac.za

28 June 2010

HREC REF: 252/2010

Ms C Scott  
 c/o Dr M Futter  
 Division of Human Genetics

Dear Ms Scott

**PROJECT TITLE: AN INVESTIGATION INTO THE EXPERIENCES OF PARENTS WITH A  
 PRE-SCHOOL CHILD WITH DOWN SYNDROME.**

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee.

It is a pleasure to inform you that the Ethics Committee has **formally approved** the above-mentioned study.

**Approval is granted for one year till the 30<sup>th</sup> June 2011.**

Please submit an annual progress report if the research continues beyond the expiry date. Please submit a brief summary of findings if you complete the study within the approval period so that we can close our file.

The reviewer congratulates the researcher on the high quality of the submission and offers the following comments for optional consideration:

**Ethics:**

1. I would like to suggest that the student consider a brief and simply worded Project Information Brochure - in addition to the Information and Consent form that the proposal supplies. This would provide more uniformity in the process of recruitment and explanation of the research (see page 11). I also think it would work better with the format of the Information and Consent form. The format as it stands asks participants to agree to a series of numbered statements. As a reader of these series of statements, I needed more information on the project itself.
2. Some of the language of the Information and Consent – as it stands – could be softened and rephrased to be more accessible to potential participants.

S Thomas